Entrez Gene Tutorial

NCBI's Entrez Gene provides gene-based information such as chromosome location, sequence, expression, structure, functional, and homology data. Each record represents a single gene from an organism. Entrez Gene includes organisms for which there is a RefSeq genome record.

In this exercise, we will learn how to obtain information about a human gene such as:

- mRNA, genomic, and protein sequence
- general gene and protein information
- homologs from other eukaryotes
- known SNPs, and whether the SNPs in the coding region alter the function of the protein product
- phenotypes associated with mutations
- protein structure

The course will also cover the advantages of Entrez Gene such as efficient searching options and availability of gene-specific information for all completely sequenced genomes, including bacteria and viruses.

The following handout includes the screen shots of Exercise 1.

Exercise 1

Retrieve human entries related to "prion protein" in Entrez Gene. Identify the gene for prion protein (PRNP). Name the map location of this gene on the human genome. What is the function of this protein? What are the alternate gene symbols? Name the phenotypes associated with the mutations in this gene.

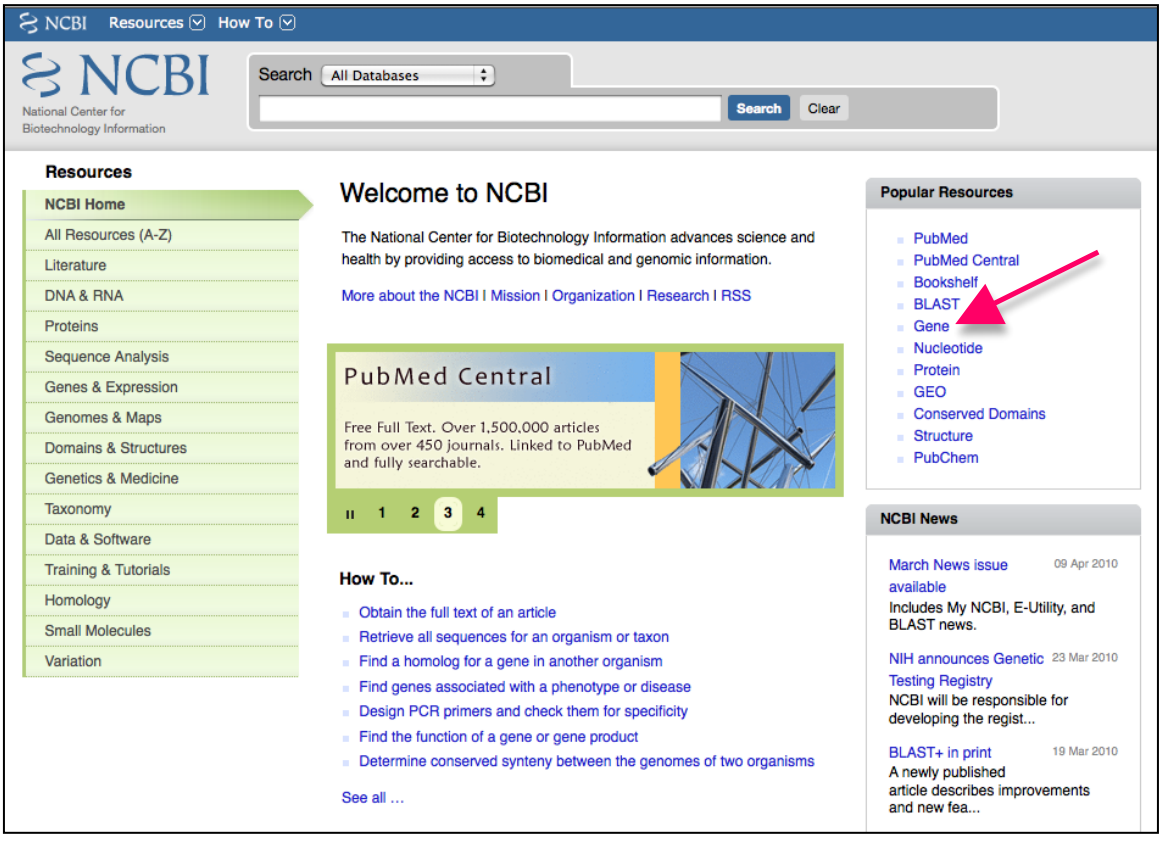
Is the RefSeq mRNA record reviewed? How many alternatively spliced products have been annotated for the gene?

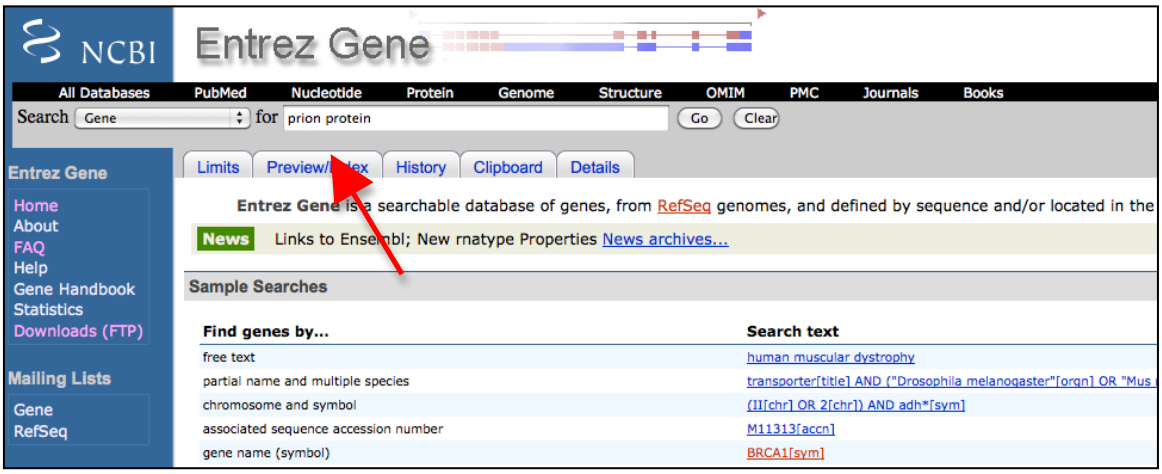
To obtain information about the homologs from other eukaryotes, click on the Homologene link. Change the Display option to "Alignment Scores". How great is the percent identity between the human and mouse proteins? View the alignment by clicking on the "Blast" link.

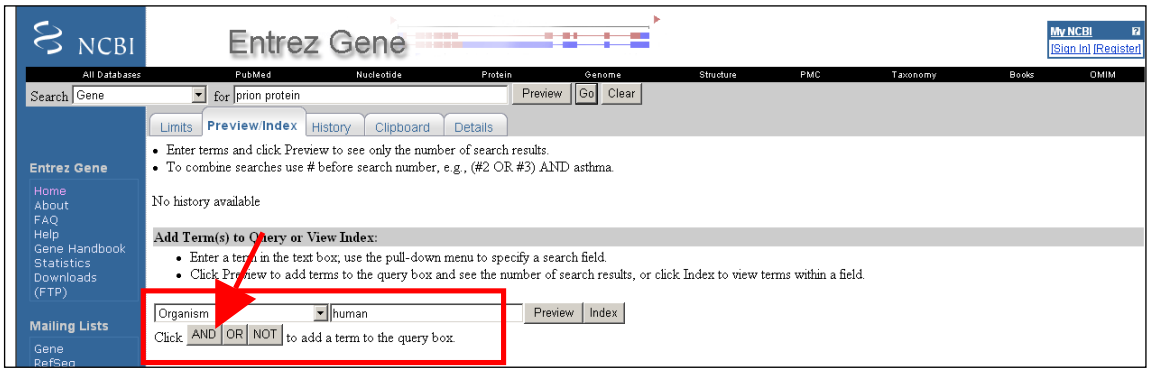
Go back to the Entrez Gene report. Identify the clinically-associated variations annotated on this gene by clicking on the SNP link. Next, Select the

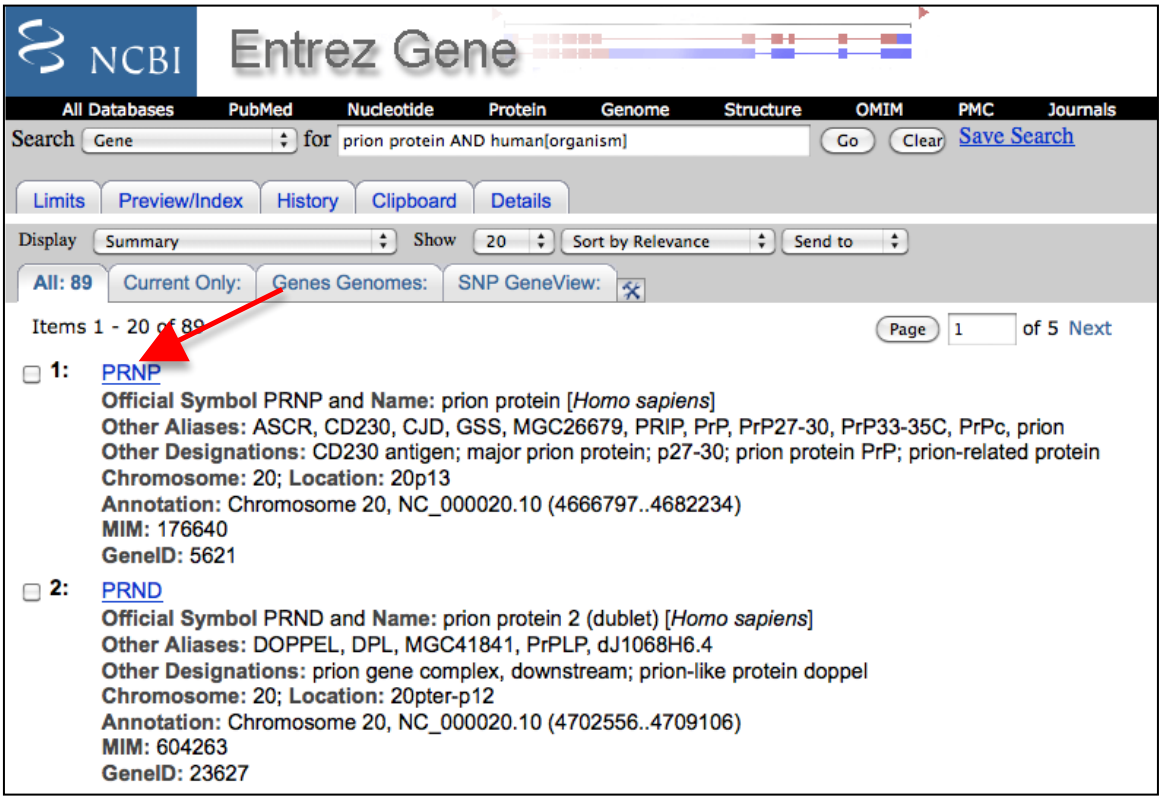
“Clinical/LSDB” tab. How many of them are missense (nonsynonymous) changes? To determine whether known SNPs in the coding region of a gene are associated with any phenotype, access the OMIM record by clicking on the icon. Compare the missense changes from the SNP report with the "ALLELIC VARIANTS" in the OMIM record. Are there any SNPs known to cause a change in the function of the prion protein?

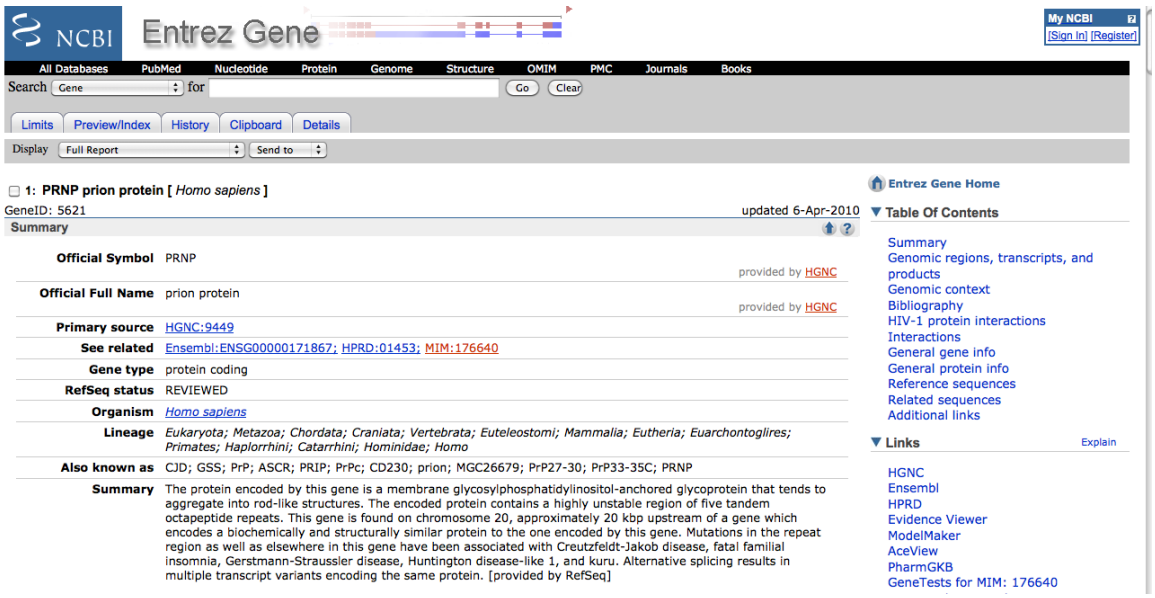
Go back to the Entrez gene report. To view the site of mutation in the 3D structure, superimpose the protein sequence on the 3D-structure of the human prion protein. Select the GENPEPT link for NP_000302 under the section “Genomic Region, Transcripts and products”. Then select “Related Structure” from the Links menu. Select “All similar MMDB” from the display drop-down menu and click Go. Next, click on the first arrow representing the related structure and then on the “Get 3D-structure data” button. Identify and highlight the residue corresponding to the mutation site the 3D structure.




NCBI
Entrez Gene
All Databases PubMed Nucleotide Protein Genome Structure OMIM PMC Journals Books
Search Gene for prion protein Go Clear
Entrez Gene
Home
About
FAQ
Help
Gene Handbook
Statistics
Downloads (FTP)
Mailing Lists
Gene
RefSeq
Limits Preview/Index History Clipboard Details
Entrez Gene is a searchable database of genes, from RefSeq genomes, and defined by sequence and/or located in the
News Links to Ensembl; New rnatype Properties News archives...
Sample Searches
Find genes by... Search text
free text human muscular dystrophy
partial name and multiple species transporter[title] AND ("Drosophila melanogaster"[orgn] OR "Mus
chromosome and symbol (II[chr] OR 2[chr]) AND adh*[sym]
associated sequence accession number M11313[accn]
gene name (symbol) BRCA1[sym]

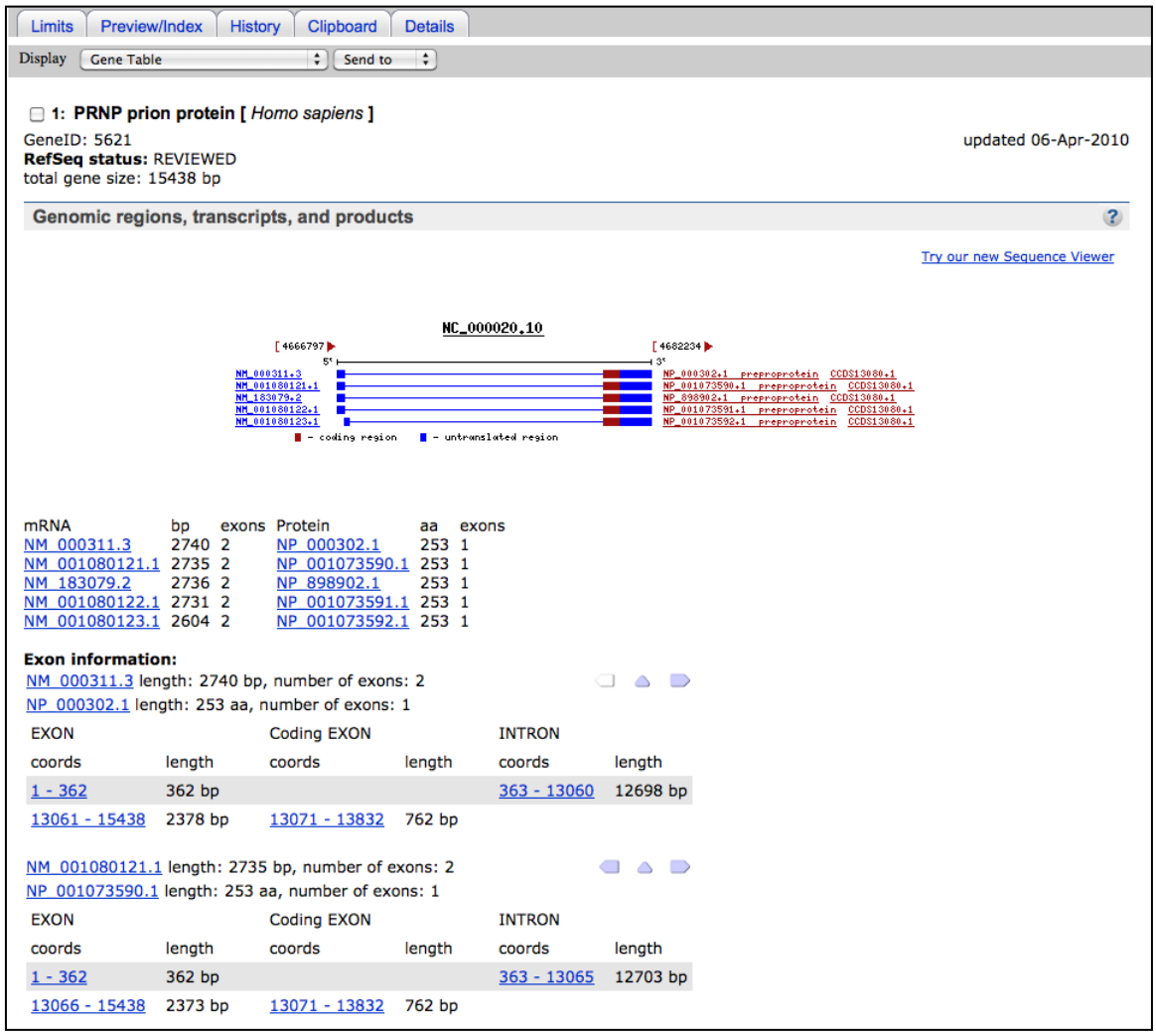

NCBI
Entrez Gene
My NCBI
[Sign In] [Register]
All Databases PubMed Nucleotide Protein Genome Structure PMC Taxonomy Books OMIM
Search Gene for prion protein Preview Go Clear
Limits Preview/Index History Clipboard Details
Entrez Gene
Home
About
FAQ
Help
Gene Handbook
Statistics
Downloads (FTP)
Mailing Lists
Gene
RefSeq
Enter terms and click Preview to see only the number of search results.
To combine searches use # before search number, e.g., (#2 OR #3) AND asthma.
No history available
Add Term(s) to Query or View Index:
Enter a term in the text box, use the pull-down menu to specify a search field.
Click Preview to add terms to the query box and see the number of search results, or click Index to view terms within a field.
Organism human Preview Index
Click AND OR NOT to add a term to the query box.


NCBI
Entrez Gene
All Databases PubMed Nucleotide Protein Genome Structure OMIM PMC Journals
Search Gene for prion protein AND human[organism] Go Clear Save Search
Limits Preview/Index History Clipboard Details
Display Summary Show 20 Sort by Relevance Send to
All: 89 Current Only: Genes Genomes: SNP GeneView:
Items 1 - 20 of 89
Page 1 of 5 Next
1: PRNP
Official Symbol PRNP and Name: prion protein [Homo sapiens]
Other Aliases: ASCR, CD230, CJD, GSS, MGC26679, PRIP, PrP, PrP27-30, PrP33-35C, PrPc, prion
Other Designations: CD230 antigen; major prion protein; p27-30; prion protein PrP; prion-related protein
Chromosome: 20; Location: 20p13
Annotation: Chromosome 20, NC_000020.10 (4666797..4682234)
MIM: 176640
GeneID: 5621
2: PRND
Official Symbol PRND and Name: prion protein 2 (dublet) [Homo sapiens]
Other Aliases: DOPPEL, DPL, MGC41841, PrPLP, dJ1068H6.4
Other Designations: prion gene complex, downstream; prion-like protein doppel
Chromosome: 20; Location: 20pter-p12
Annotation: Chromosome 20, NC_000020.10 (4702556..4709106)
MIM: 604263
GeneID: 23627


NCBI
Entrez Gene
My NCBI [Sign In] [Register]
All Databases PubMed Nucleotide Protein Genome Structure OMIM PMC Journals Books
Search Gene for Go Clear
Limits Preview/Index History Clipboard Details
Display Full Report Send to
1: PRNP prion protein [Homo sapiens]
GeneID: 5621 updated 6-Apr-2010
Summary
Official Symbol PRNP provided by HGNC
Official Full Name prion protein provided by HGNC
Primary source HGNC:9449
See related Ensembl:ENSG00000171867; HPRD:01453; MIM:176640
Gene type protein coding
RefSeq status REVIEWED
Organism Homo sapiens
Lineage Eukaryota; Metazoa; Chordata; Craniata; Vertebrata; Euteleostomi; Mammalia; Eutheria; Euarchontoglires; Primates; Haplorrhini; Catarrhini; Hominidae; Homo
Also known as CJD; GSS; PrP; ASCR; PRIP; PrPc; CD230; prion; MGC26679; PrP27-30; PrP33-35C; PRNP
Summary The protein encoded by this gene is a membrane glycosylphosphatidylinositol-anchored glycoprotein that tends to aggregate into rod-like structures. The encoded protein contains a highly unstable region of five tandem octapeptide repeats. This gene is found on chromosome 20, approximately 20 kbp upstream of a gene which encodes a biochemically and structurally similar protein to the one encoded by this gene. Mutations in the repeat region as well as elsewhere in this gene have been associated with Creutzfeldt-Jakob disease, fatal familial insomnia, Gerstmann-Straussler disease, Huntington disease-like 1, and kuru. Alternative splicing results in multiple transcript variants encoding the same protein. [provided by RefSeq]
Entrez Gene Home
Table Of Contents
Summary
Genomic regions, transcripts, and products
Genomic context
Bibliography
HIV-1 protein interactions
Interactions
General gene info
General protein info
Reference sequences
Related sequences
Additional links
Links Explain
HGNC
Ensembl
HPRD
Evidence Viewer
ModelMaker
AceView
PharmGKB
GeneTests for MIM: 176640


Genomic regions, transcripts, and products
Go to reference sequence details
Try our new Sequence Viewer
NC_000020.9
Genomic context
chromosome: 20; Location: 20p13
See PRNP in MapViewer
SNP: Genotype
SNP: GeneView
Taxonomy
UniSTS
AceView
CCDS
Ensembl
Evidence Viewer
GDB
GeneTests for MIM: 176640
HGMD
HGNC
HPRD
HuGE Navigator
KEGG
MGC
ModelMaker
PharmGKB
Prion Protein/CJD database
UniGene
LinkOut
Entrez Gene Info
Feedback
Subscriptions


NCBI
Entrez Gene
My NCBI
[Sign In] [Register]
All Databases
PubMed
Nucleotide
Protein
Genome
Structure
PMC
Taxonomy
Books
OMIM
Search Gene for
Go
Clear
Limits
Preview/Index
History
Clipboard
Details
Display Full Report
Show 20
Send to
All: 1
SNP GeneView: 1
Full Report
Summary
Brief
ASN.1
XML
Gene Table
UI List
LinkOut
Books Links
Conserved Domain Links
Genome Links
GENSAT Links
GEO Profile Links
HomoloGene Links
CoreNucleotide Links
CoreNucleotide NIH cDNA clone links
EST Links
EST NIH cDNA clone links
GSS Links
GSS NIH cDNA clone links
Nucleotide Links
NIH cDNA clone links
OMIA Links
OMIM Links
BioAssay Links
PubChem Compound Links
PubChem Substance Links
PMC Links
Probe Links
Protein Links
tzfeldt-Jakob disease, Gerstmann-Strausler-Scheinker syndrome, fatal familial insomnia)
updated 28-Oct-2007
provided by HGNC
30) (Creutzfeldt-Jakob disease, Gerstmann-Strausler-Scheinker syndrome, fatal familial
provided by HGNC
0171867; HPRD:01453; MIM:176640
a; Chordata; Craniata; Vertebrata; Euteleostomi; Mammalia; Eutheria; Euarchontoglires;
ini; Catarrhini; Hominidae; Homo
CR; PRIP; PrPc; CD230; MGC26679; PrP27-30; PrP33-35C
ed by this gene is a membrane glycosylphosphatidylinositol-anchored glycoprotein that tends to
-like structures. The encoded protein contains a highly unstable region of five tandem
s. This gene is found on chromosome 20, approximately 20 kbp upstream of a gene which
nically and structurally similar protein to the one encoded by this gene. Mutations in the repeat
region as well as elsewhere in this gene have been associated with Creutzfeldt-Jakob disease, fatal familial
Entrez Gene Home
Table Of Contents
Summary
Genomic regions, transcripts...
Genomic context
Bibliography
HIV-1 protein interactions
Interactions
General gene information
General protein information
Reference Sequences
Related Sequences
Additional Links
Links
Explain
Order cDNA clone
Books
Conserved Domains
Genome
GEO Profiles
HomoloGene
Map Viewer
CoreNucleotide
EST
Nucleotide
OMIA
OMIM



Limits | Preview/Index | History | Clipboard | Details

Display: Gene Table | Send to

1: PRNP prion protein [*Homo sapiens*]

GeneID: 5621 — updated 06-Apr-2010

RefSeq status: REVIEWED

total gene size: 15438 bp

Genomic regions, transcripts, and products

Try our new Sequence Viewer

mRNA	bp	exons	Protein	aa	exons
NM_000311.3	2740	2	NP_000302.1	253	1
NM_001080121.1	2735	2	NP_001073590.1	253	1
NM_183079.2	2736	2	NP_898902.1	253	1
NM_001080122.1	2731	2	NP_001073591.1	253	1
NM_001080123.1	2604	2	NP_001073592.1	253	1

Exon information:

NM_000311.3 length: 2740 bp, number of exons: 2

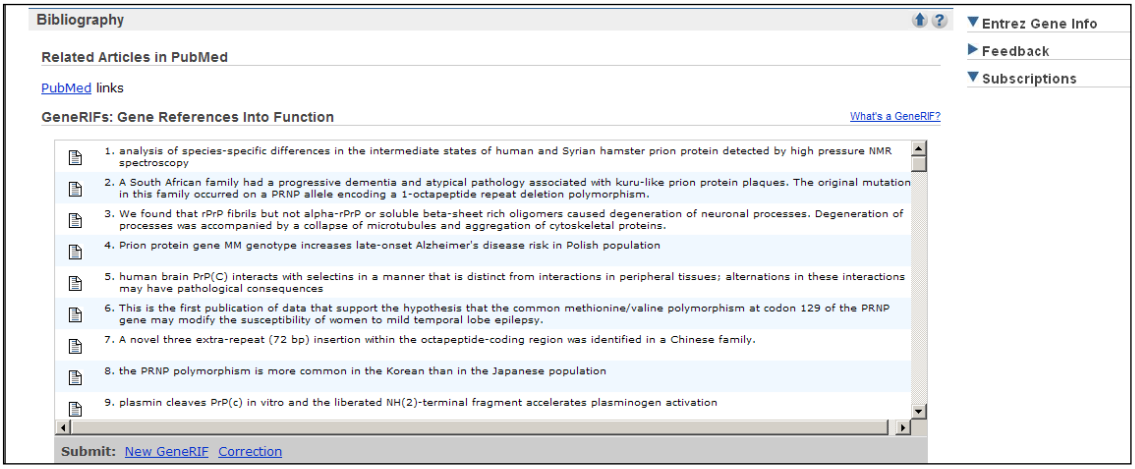
NP_000302.1 length: 253 aa, number of exons: 1

EXON		Coding EXON		INTRON	
coords	length	coords	length	coords	length
1 - 362	362 bp			363 - 13060	12698 bp
13061 - 15438	2378 bp	13071 - 13832	762 bp		

NM_001080121.1 length: 2735 bp, number of exons: 2

NP_001073590.1 length: 253 aa, number of exons: 1

EXON		Coding EXON		INTRON	
coords	length	coords	length	coords	length
1 - 362	362 bp			363 - 13065	12703 bp
13066 - 15438	2373 bp	13071 - 13832	762 bp		



Bibliography

Related Articles in PubMed

PubMed links

GeneRIFs: Gene References Into Function — What's a GeneRIF?

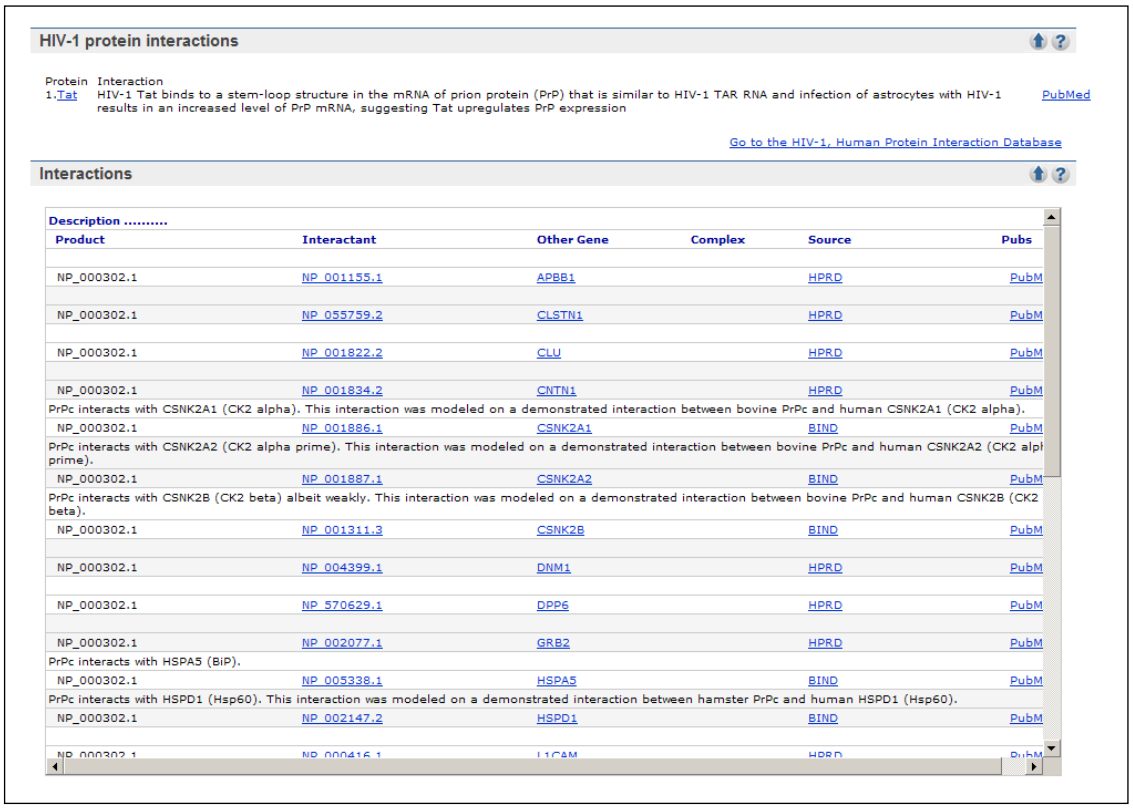
1. analysis of species-specific differences in the intermediate states of human and Syrian hamster prion protein detected by high pressure NMR spectroscopy
2. A South African family had a progressive dementia and atypical pathology associated with kuru-like prion protein plaques. The original mutation in this family occurred on a PRNP allele encoding a 1-octapeptide repeat deletion polymorphism.
3. We found that rPrP fibrils but not alpha-rPrP or soluble beta-sheet rich oligomers caused degeneration of neuronal processes. Degeneration of processes was accompanied by a collapse of microtubules and aggregation of cytoskeletal proteins.
4. Prion protein gene MM genotype increases late-onset Alzheimer's disease risk in Polish population
5. human brain PrP(C) interacts with selectins in a manner that is distinct from interactions in peripheral tissues; alternations in these interactions may have pathological consequences
6. This is the first publication of data that support the hypothesis that the common methionine/valine polymorphism at codon 129 of the PRNP gene may modify the susceptibility of women to mild temporal lobe epilepsy.
7. A novel three extra-repeat (72 bp) insertion within the octapeptide-coding region was identified in a Chinese family.
8. the PRNP polymorphism is more common in the Korean than in the Japanese population
9. plasmin cleaves PrP(c) in vitro and the liberated NH(2)-terminal fragment accelerates plasminogen activation

Submit: New GeneRIF Correction

Entrez Gene Info

Feedback

Subscriptions



HIV-1 protein interactions

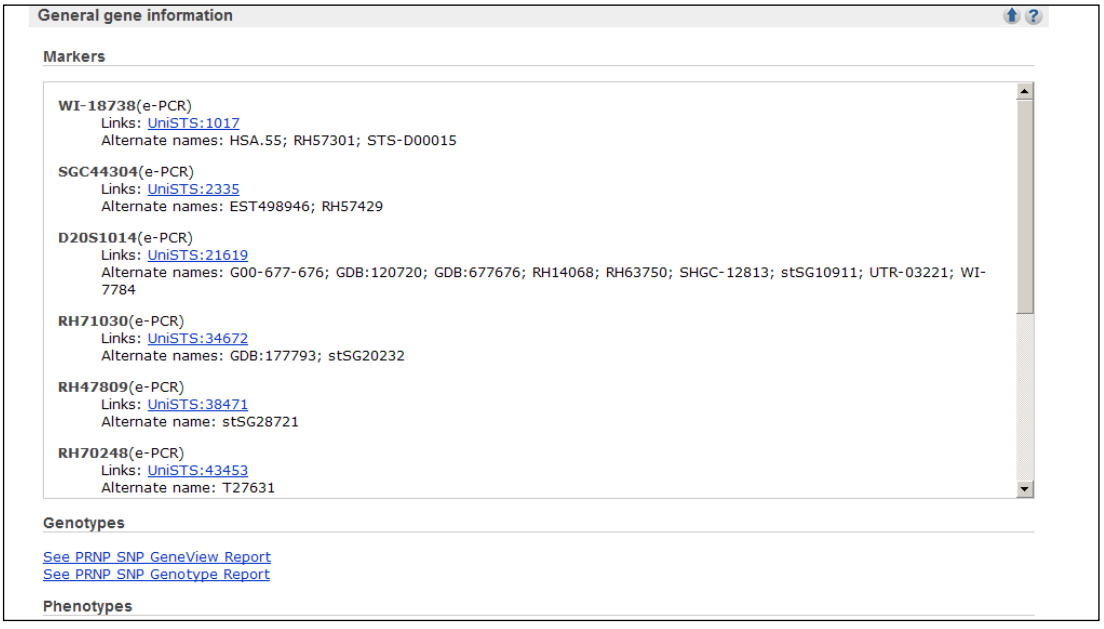
Protein Interaction
1.Tat HIV-1 Tat binds to a stem-loop structure in the mRNA of prion protein (PrP) that is similar to HIV-1 TAR RNA and infection of astrocytes with HIV-1 results in an increased level of PrP mRNA, suggesting Tat upregulates PrP expression PubMed

Go to the HIV-1, Human Protein Interaction Database

Interactions

Description .....

Product	Interactant	Other Gene	Complex	Source	Pubs
NP_000302.1	NP_001155.1	APBB1		HPRD	PubM
NP_000302.1	NP_055759.2	CLSTN1		HPRD	PubM
NP_000302.1	NP_001822.2	CLU		HPRD	PubM
NP_000302.1	NP_001834.2	CNTN1		HPRD	PubM
PrPc interacts with CSNK2A1 (CK2 alpha). This interaction was modeled on a demonstrated interaction between bovine PrPc and human CSNK2A1 (CK2 alpha).					
NP_000302.1	NP_001886.1	CSNK2A1		BIND	PubM
PrPc interacts with CSNK2A2 (CK2 alpha prime). This interaction was modeled on a demonstrated interaction between bovine PrPc and human CSNK2A2 (CK2 alpha prime).					
NP_000302.1	NP_001887.1	CSNK2A2		BIND	PubM
PrPc interacts with CSNK2B (CK2 beta) albeit weakly. This interaction was modeled on a demonstrated interaction between bovine PrPc and human CSNK2B (CK2 beta).					
NP_000302.1	NP_001311.3	CSNK2B		BIND	PubM
NP_000302.1	NP_004399.1	DNM1		HPRD	PubM
NP_000302.1	NP_570629.1	DPP6		HPRD	PubM
NP_000302.1	NP_002077.1	GRB2		HPRD	PubM
PrPc interacts with HSPA5 (BiP).					
NP_000302.1	NP_005338.1	HSPA5		BIND	PubM
PrPc interacts with HSPD1 (Hsp60). This interaction was modeled on a demonstrated interaction between hamster PrPc and human HSPD1 (Hsp60).					
NP_000302.1	NP_002147.2	HSPD1		BIND	PubM
NP_000302.1	NP_000416.1	L1CAM		HPRD	PubM



General gene information

Markers

WI-18738(e-PCR)
Links: UniSTS:1017
Alternate names: HSA.55; RH57301; STS-D00015

SGC44304(e-PCR)
Links: UniSTS:2335
Alternate names: EST498946; RH57429

D20S1014(e-PCR)
Links: UniSTS:21619
Alternate names: G00-677-676; GDB:120720; GDB:677676; RH14068; RH63750; SHGC-12813; stSG10911; UTR-03221; WI-7784

RH71030(e-PCR)
Links: UniSTS:34672
Alternate names: GDB:177793; stSG20232

RH47809(e-PCR)
Links: UniSTS:38471
Alternate name: stSG28721

RH70248(e-PCR)
Links: UniSTS:43453
Alternate name: T27631

Genotypes

See PRNP SNP GeneView Report
See PRNP SNP Genotype Report

Phenotypes

Phenotypes

Creutzfeldt-Jakob disease
MIM: 123400
Gerstmann-Straussler disease
MIM: 137440
Huntington disease-like 1
MIM: 603218
Insomnia, fatal familial
MIM: 600072
Prion disease with protracted course
MIM: 606688

Pathways

KEGG pathway: Neurodegenerative Disorders
01510
KEGG pathway: Prion disease
05060

Homology

Mouse, Rat
Map Viewer

GeneOntology

Provided by GOA

Function	Evidence	
GPI anchor binding	IEA	
copper ion binding	TAS	PubMed
microtubule binding	IDA	PubMed
protein binding	IPI	PubMed

Process	Evidence	
cellular copper ion homeostasis	NAS	PubMed
metabolic process	TAS	PubMed
response to oxidative stress	ISS	

Component	Evidence	
Golgi apparatus	ISS	
cytoplasm	TAS	PubMed
endoplasmic reticulum	ISS	
extrinsic to membrane	TAS	PubMed
lipid raft	ISS	
membrane	IEA	
plasma membrane	ISS	

General protein information

General protein information

Names

prion protein
CD230 antigen
prion protein PrP
major prion protein
prion-related protein

NCBI Reference Sequences (RefSeq)

RefSeqs maintained independently of Annotated Genomes

These reference sequences exist independently of genome builds. Explain

mRNA and Protein(s)

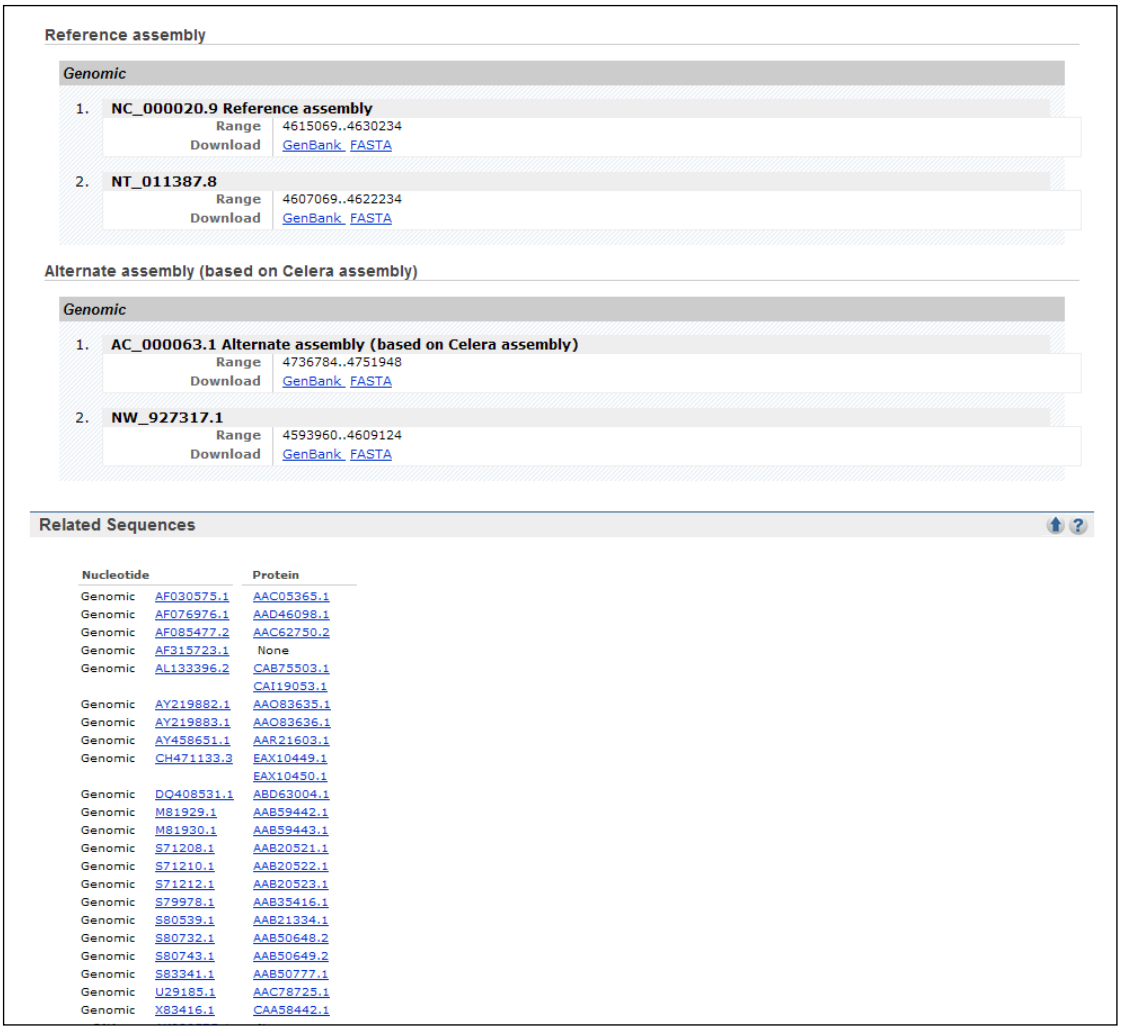
1. **NM_000311.3→NP_000302.1 prion protein preproprotein**
   - Description: Transcript Variant: This variant (1) represents the longest transcript. Variants 1-5 encode the same protein.
   - Source sequence(s): AW452130,BC022532,DA297032,M13899
   - Consensus CDS: CCDS13080.1
   - Conserved Domains (1) summary
     - smart00157 Location:23–230 Blast Score:546: PRP; Major prion protein; The prion protein is a major component of scrapie-associated fibrils in Creutzfeldt-Jakob disease, kuru, Gerstmann-Straussler syndrome and bovine spongiform encephalopathy.

2. **NM_001080121.1→NP_001073590.1 prion protein preproprotein**
   - Description: Transcript Variant: This variant (3) uses an alternate splice site in the 5' UTR compared to variant 1. Variants 1-5 encode the same protein.
   - Source sequence(s): AW452130,BC022532,BP251427,DA122620,M13899
   - Conserved Domains (1) summary
     - smart00157 Location:23–230 Blast Score:546: PRP; Major prion protein; The prion protein is a major component of scrapie-associated fibrils in Creutzfeldt-Jakob disease, kuru, Gerstmann-Straussler syndrome and bovine spongiform encephalopathy.

3. **NM_001080122.1→NP_001073591.1 prion protein preproprotein**
   - Description: Transcript Variant: This variant (4) uses an alternate splice site in the 5' UTR compared to variant 1. Variants 1-5 encode the same protein.
   - Source sequence(s): AW452130,BC022532,BI669189,DA297032,M13899
   - Conserved Domains (1) summary
     - smart00157 Location:23–230 Blast Score:546: PRP; Major prion protein; The prion protein is a major component of scrapie-associated fibrils in Creutzfeldt-Jakob disease, kuru, Gerstmann-Straussler syndrome and bovine spongiform encephalopathy.

4. **NM_001080123.1→NP_001073592.1 prion protein preproprotein**
   - Description: Transcript Variant: This variant (5) uses an alternate splice site in the 5' UTR compared to variant 1. Variants 1-5 encode the same protein.
   - Source sequence(s): BC022532,DB461478,M13899
   - Conserved Domains (1) summary
     - smart00157 Location:23–230 Blast Score:546: PRP; Major prion protein; The prion protein is a major component of scrapie-associated fibrils in Creutzfeldt-Jakob disease, kuru, Gerstmann-Straussler syndrome and bovine spongiform encephalopathy.

5. **NM_183079.2→NP_898902.1 prion protein preproprotein**
   - Description: Transcript Variant: This variant (2) uses an alternate splice site in the 5' UTR compared to variant 1. Variants 1-5 encode the same protein.
   - Source sequence(s): AW452130,AY008282,BC022532,DA122620
   - Consensus CDS: CCDS13080.1
   - Conserved Domains (1) summary
     - smart00157: PRP; Major prion protein; The prion protein is a major component of scrapie-associated fibrils in



Reference assembly

Genomic

1. NC_000020.9 Reference assembly
   Range 4615069..4630234
   Download GenBank FASTA
2. NT_011387.8
   Range 4607069..4622234
   Download GenBank FASTA

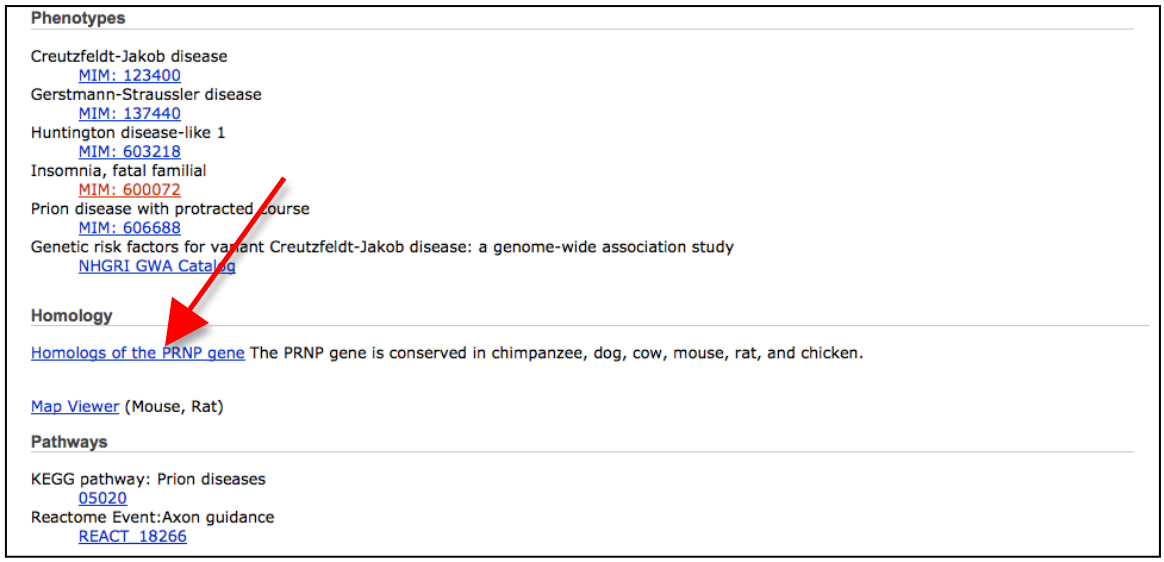
Alternate assembly (based on Celera assembly)

Genomic

1. AC_000063.1 Alternate assembly (based on Celera assembly)
   Range 4736784..4751948
   Download GenBank FASTA
2. NW_927317.1
   Range 4593960..4609124
   Download GenBank FASTA

Related Sequences

Nucleotide		Protein
Genomic	AF030575.1	AAC05365.1
Genomic	AF076976.1	AAD46098.1
Genomic	AF085477.2	AAC62750.2
Genomic	AF315723.1	None
Genomic	AL133396.2	CAB75503.1
		CAI19053.1
Genomic	AY219882.1	AAO83635.1
Genomic	AY219883.1	AAO83636.1
Genomic	AY458651.1	AAR21603.1
Genomic	CH471133.3	EAX10449.1
		EAX10450.1
Genomic	DQ408531.1	ABD63004.1
Genomic	M81929.1	AAB59442.1
Genomic	M81930.1	AAB59443.1
Genomic	S71208.1	AAB20521.1
Genomic	S71210.1	AAB20522.1
Genomic	S71212.1	AAB20523.1
Genomic	S79978.1	AAB35416.1
Genomic	S80539.1	AAB21334.1
Genomic	S80732.1	AAB50648.2
Genomic	S80743.1	AAB50649.2
Genomic	S83341.1	AAB50777.1
Genomic	U29185.1	AAC78725.1
Genomic	X83416.1	CAA58442.1



Phenotypes

Creutzfeldt-Jakob disease
MIM: 123400
Gerstmann-Straussler disease
MIM: 137440
Huntington disease-like 1
MIM: 603218
Insomnia, fatal familial
MIM: 600072
Prion disease with protracted course
MIM: 606688
Genetic risk factors for variant Creutzfeldt-Jakob disease: a genome-wide association study
NHGRI GWA Catalog

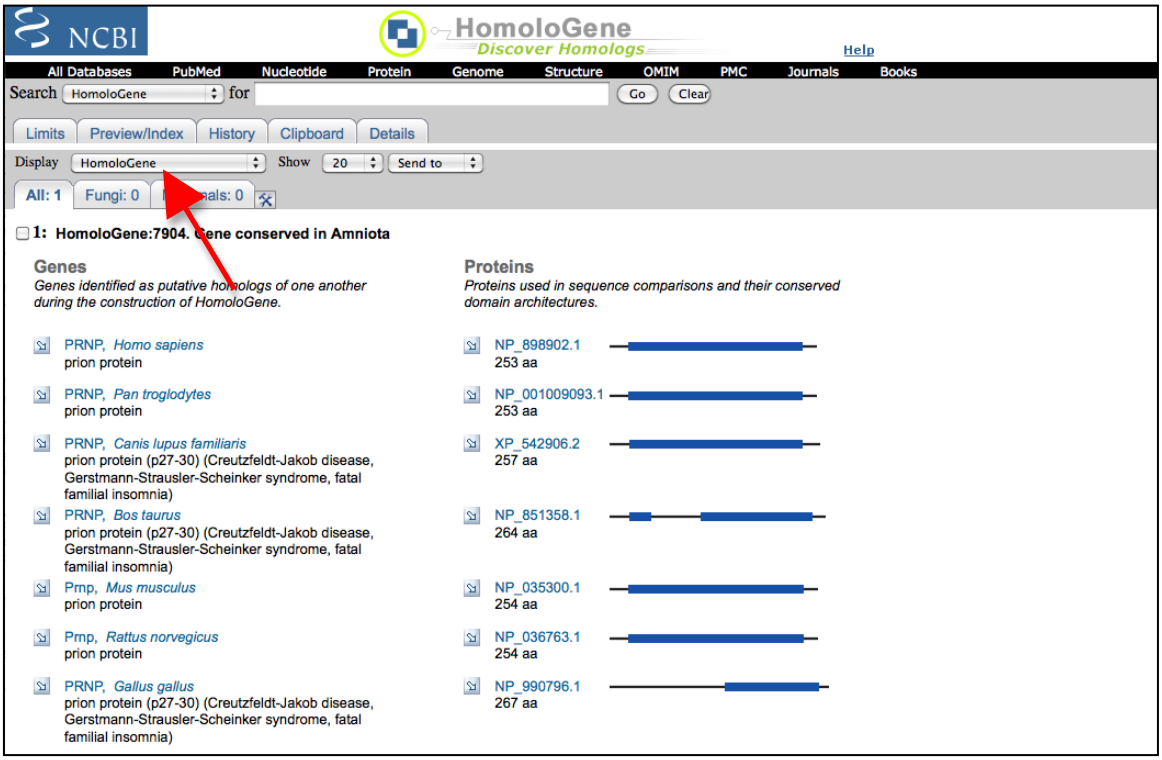
Homology

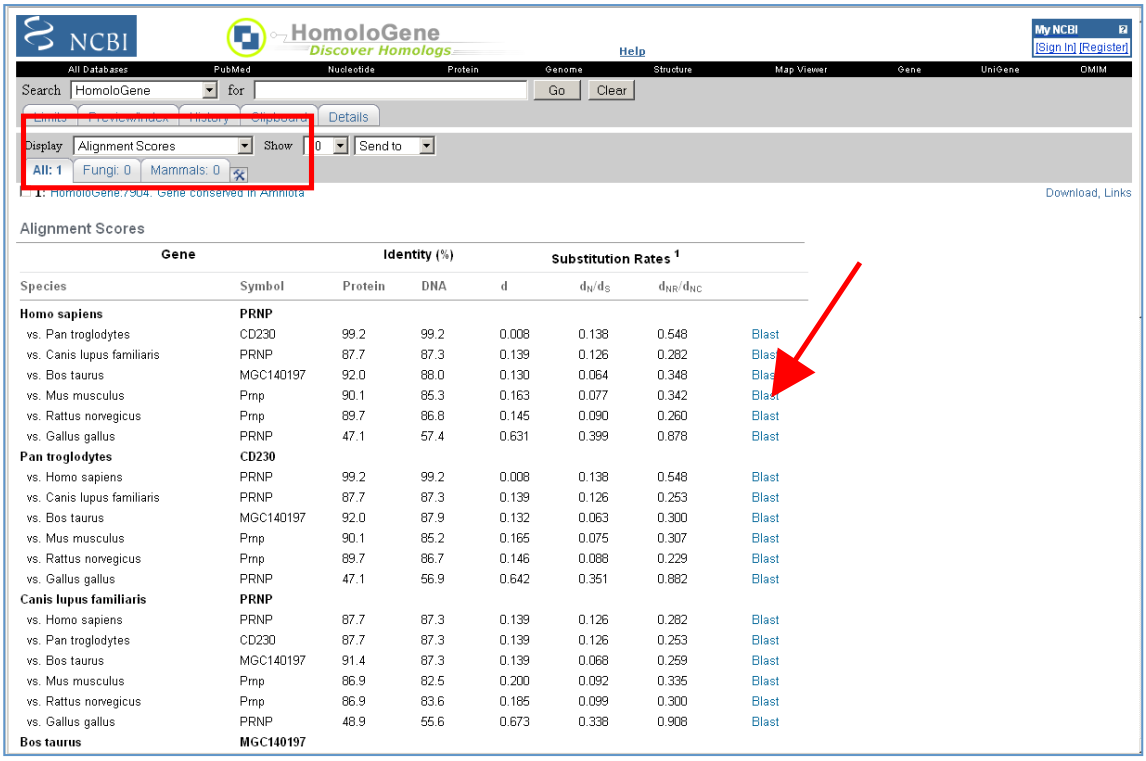
Homologs of the PRNP gene The PRNP gene is conserved in chimpanzee, dog, cow, mouse, rat, and chicken.

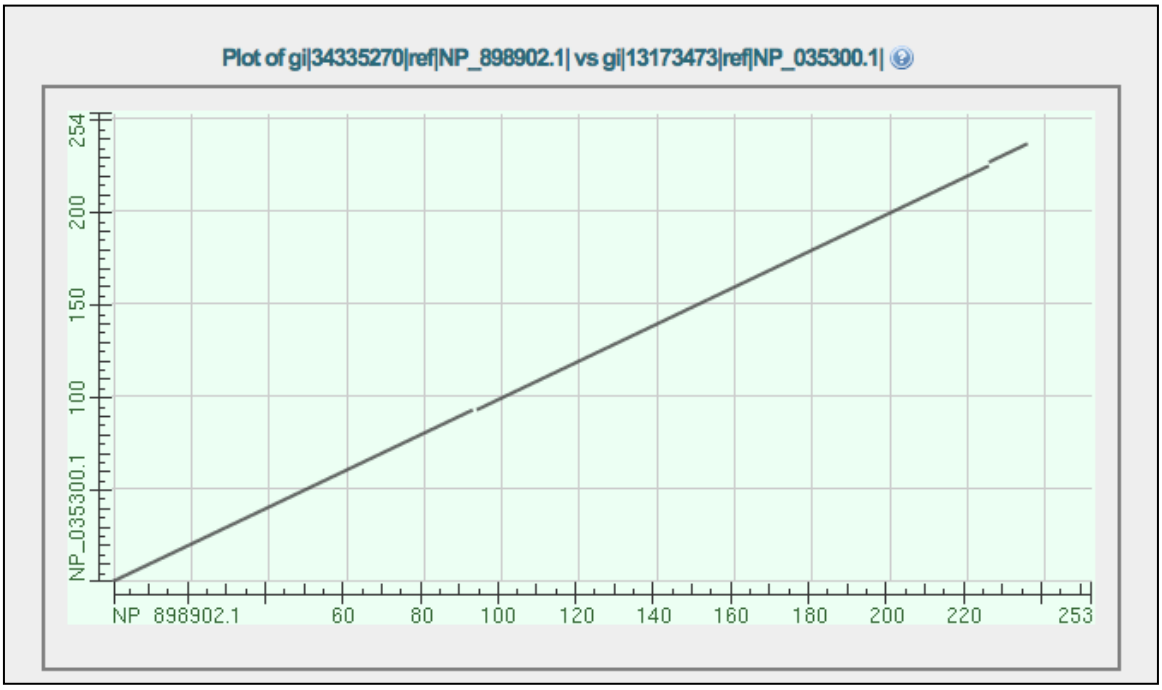
Map Viewer (Mouse, Rat)

Pathways

KEGG pathway: Prion diseases
05020
Reactome Event:Axon guidance
REACT_18266


NCBI
HomoloGene
Discover Homologs
Help
All Databases PubMed Nucleotide Protein Genome Structure OMIM PMC Journals Books
Search HomoloGene for Go Clear
Limits Preview/Index History Clipboard Details
Display HomoloGene Show 20 Send to
All: 1 Fungi: 0
1: HomoloGene:7904. Gene conserved in Amniota
Genes
Genes identified as putative homologs of one another during the construction of HomoloGene.
Proteins
Proteins used in sequence comparisons and their conserved domain architectures.
PRNP, Homo sapiens
prion protein
NP_898902.1
253 aa
PRNP, Pan troglodytes
prion protein
NP_001009093.1
253 aa
PRNP, Canis lupus familiaris
prion protein (p27-30) (Creutzfeldt-Jakob disease, Gerstmann-Strausler-Scheinker syndrome, fatal familial insomnia)
XP_542906.2
257 aa
PRNP, Bos taurus
prion protein (p27-30) (Creutzfeldt-Jakob disease, Gerstmann-Strausler-Scheinker syndrome, fatal familial insomnia)
NP_851358.1
264 aa
Prnp, Mus musculus
prion protein
NP_035300.1
254 aa
Prnp, Rattus norvegicus
prion protein
NP_036763.1
254 aa
PRNP, Gallus gallus
prion protein (p27-30) (Creutzfeldt-Jakob disease, Gerstmann-Strausler-Scheinker syndrome, fatal familial insomnia)
NP_990796.1
267 aa


NCBI
HomoloGene
Discover Homologs
Help
My NCBI [Sign In] [Register]
All Databases PubMed Nucleotide Protein Genome Structure Map Viewer Gene UniGene OMIM
Search HomoloGene for Go Clear
Limits Preview/Index History Clipboard Details
Display Alignment Scores Show 0 Send to
All: 1 Fungi: 0 Mammals: 0
1: HomoloGene:7904. Gene conserved in Amniota
Download, Links
Alignment Scores
Gene
Identity (%)
Substitution Rates 1
Species Symbol Protein DNA d dN/dS dNR/dNC
Homo sapiens PRNP
vs. Pan troglodytes CD230 99.2 99.2 0.008 0.138 0.548 Blast
vs. Canis lupus familiaris PRNP 87.7 87.3 0.139 0.126 0.282 Blast
vs. Bos taurus MGC140197 92.0 88.0 0.130 0.064 0.348 Blast
vs. Mus musculus Prnp 90.1 85.3 0.163 0.077 0.342 Blast
vs. Rattus norvegicus Prnp 89.7 86.8 0.145 0.090 0.260 Blast
vs. Gallus gallus PRNP 47.1 57.4 0.631 0.399 0.878 Blast
Pan troglodytes CD230
vs. Homo sapiens PRNP 99.2 99.2 0.008 0.138 0.548 Blast
vs. Canis lupus familiaris PRNP 87.7 87.3 0.139 0.126 0.253 Blast
vs. Bos taurus MGC140197 92.0 87.9 0.132 0.063 0.300 Blast
vs. Mus musculus Prnp 90.1 85.2 0.165 0.075 0.307 Blast
vs. Rattus norvegicus Prnp 89.7 86.7 0.146 0.088 0.229 Blast
vs. Gallus gallus PRNP 47.1 56.9 0.642 0.351 0.882 Blast
Canis lupus familiaris PRNP
vs. Homo sapiens PRNP 87.7 87.3 0.139 0.126 0.282 Blast
vs. Pan troglodytes CD230 87.7 87.3 0.139 0.126 0.253 Blast
vs. Bos taurus MGC140197 91.4 87.3 0.139 0.068 0.259 Blast
vs. Mus musculus Prnp 86.9 82.5 0.200 0.092 0.335 Blast
vs. Rattus norvegicus Prnp 86.9 83.6 0.185 0.099 0.300 Blast
vs. Gallus gallus PRNP 48.9 55.6 0.673 0.338 0.908 Blast
Bos taurus MGC140197



Descriptions

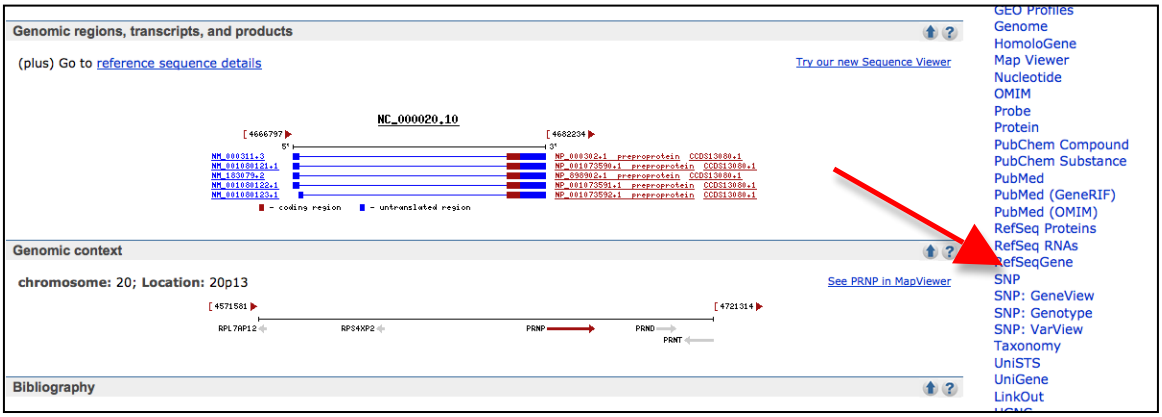
Sequences producing significant alignments:	Score (Bits)	E Value
ref|NP_035300.1| major prion protein precursor [Mus musculus]...	252	5e-72

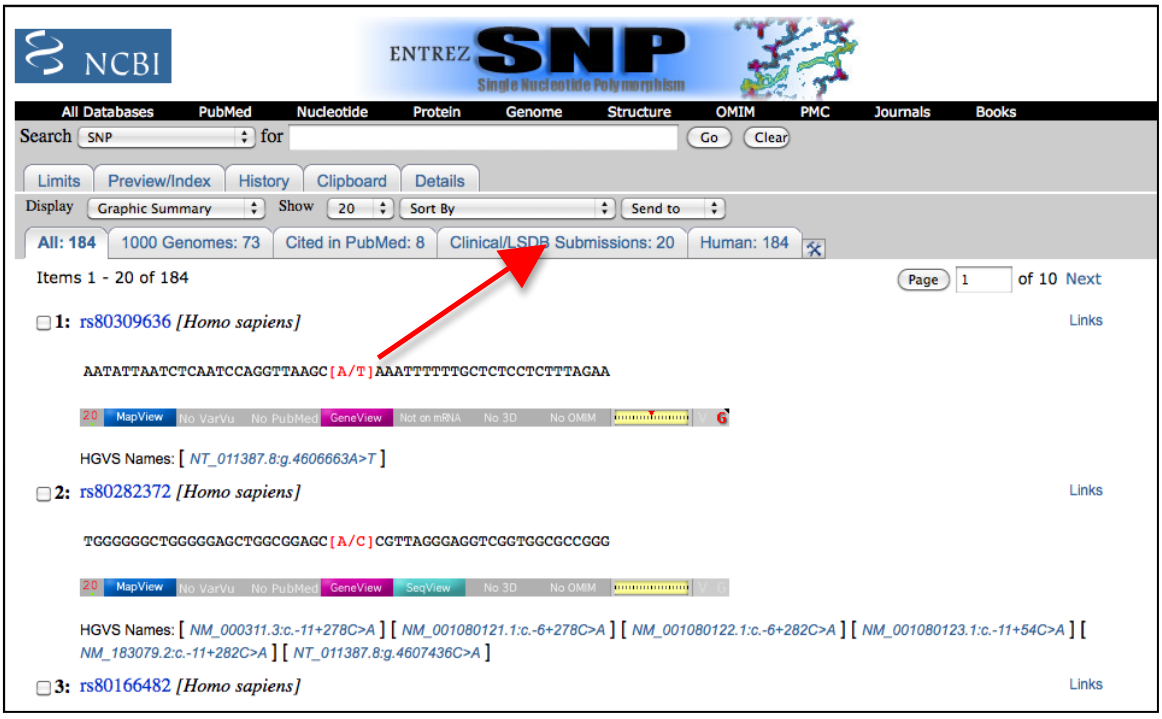
Alignments

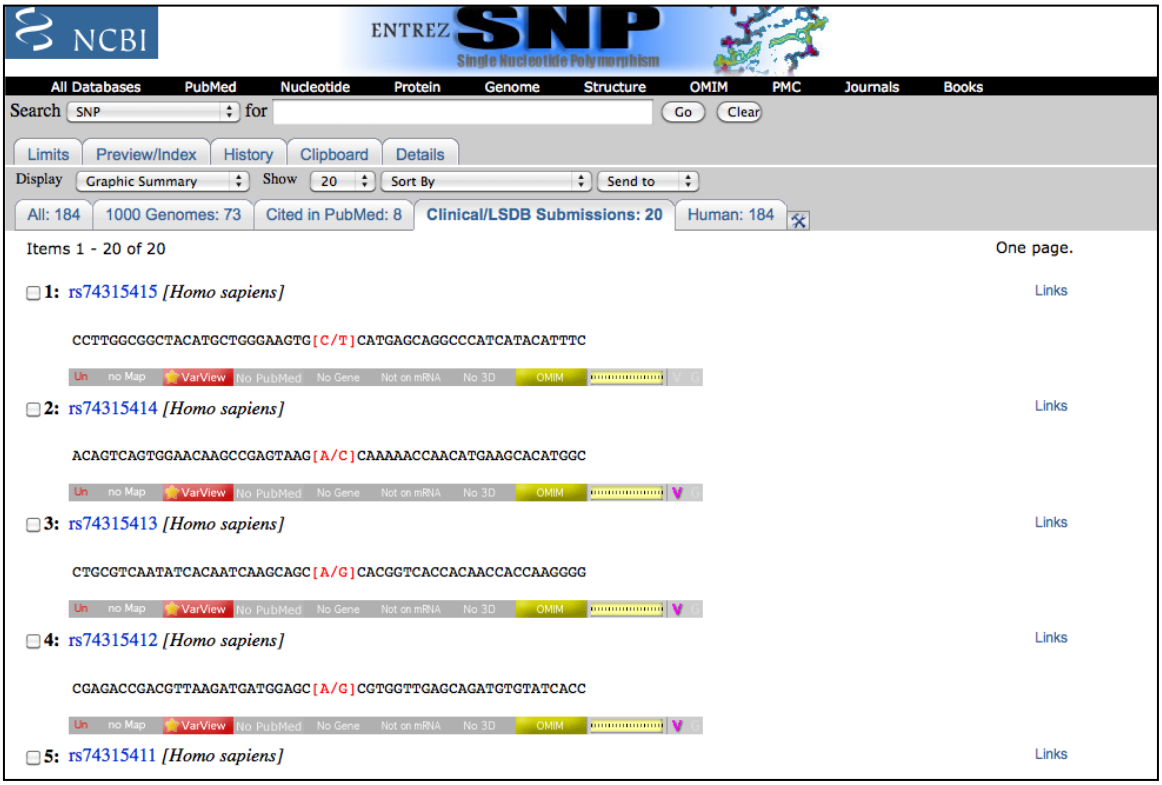
>ref|NP_035300.1| major prion protein precursor [Mus musculus]
sp|P04925.2|PRIO_MOUSE RecName: Full=Major prion protein; Short=PrP; AltName: Full=PrP27-30;
AltName: Full=PrP33-35C; AltName: CD_antigen=CD230;
Flags: Precursor
gb|AAA39997.1| prion protein [Mus musculus]
▷9 more sequence titles
Length=254

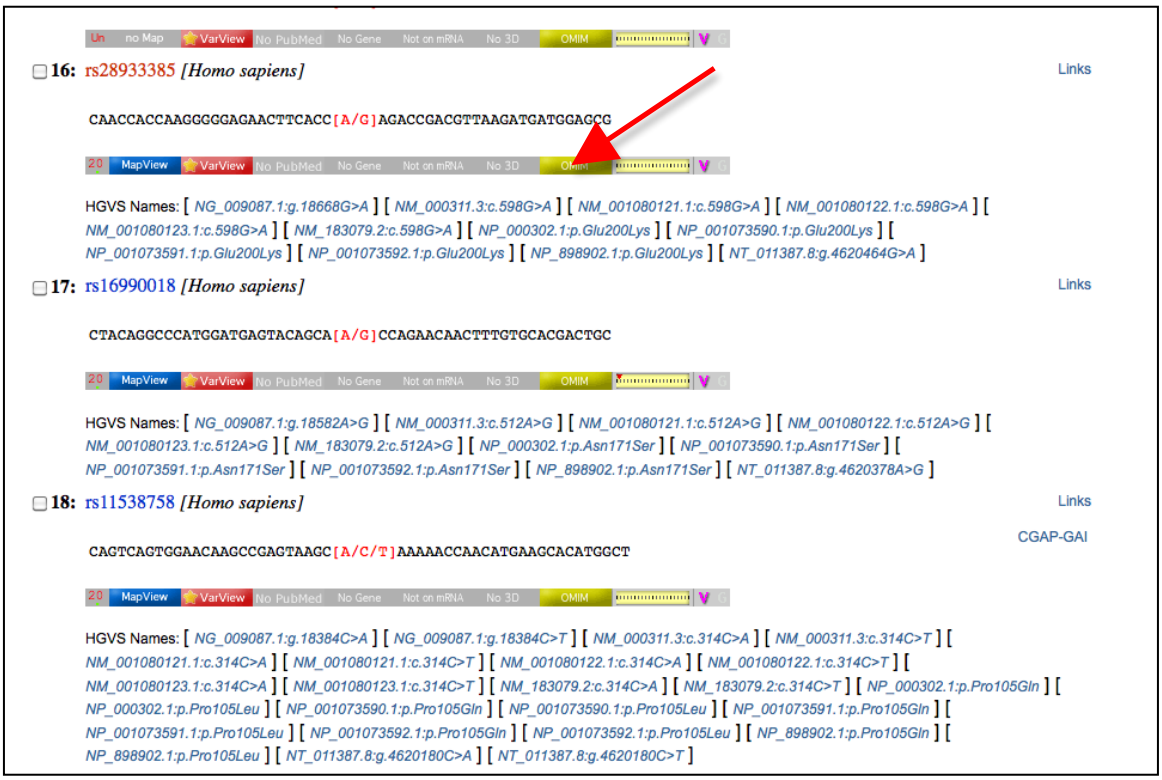
Score = 252 bits (644), Expect = 5e-72, Method: Compositional matrix adjust.
Identities = 182/238 (76%), Positives = 198/238 (83%), Gaps = 3/238 (1%)

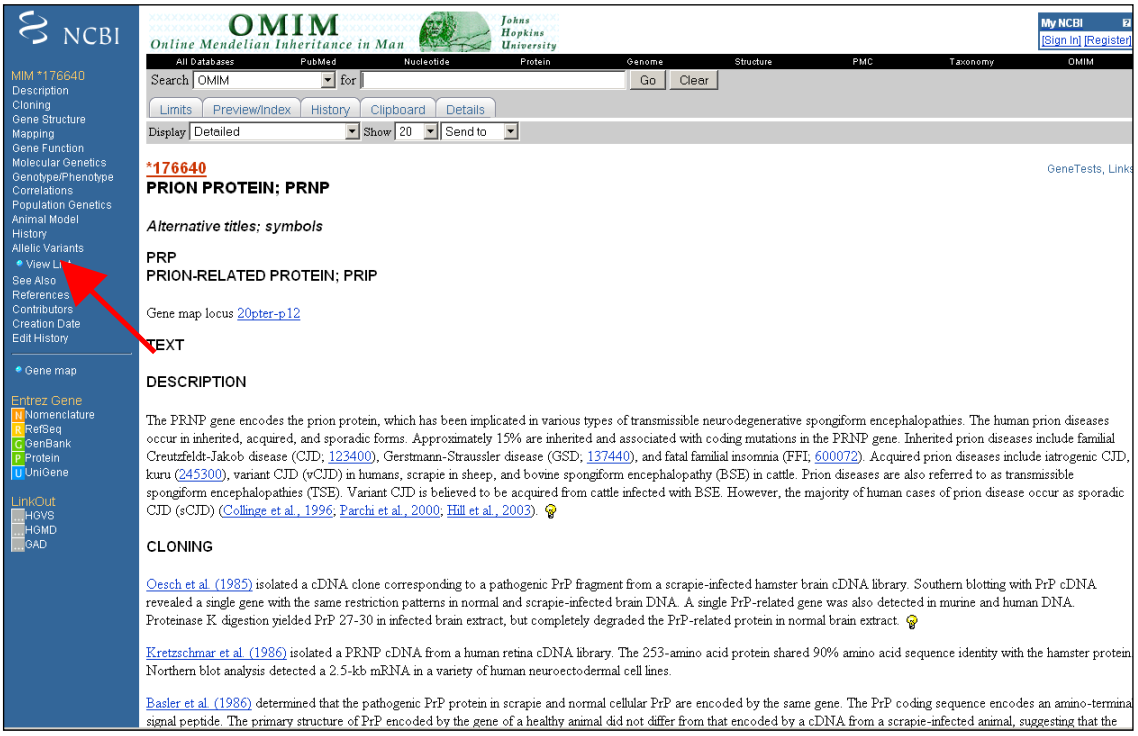
```
Query  1    MANLGCWMLVLFVATWSDLGLCKKRPKPGGWNTGGSRYPGQGSPGGNRYppqggggwgqp  60
            MANLG W+L LFV  W+D+GLCKKRPKPGGWNTGGSRYPGQGSPGGNRYPPQGG
Sbjct  1    MANLGYWLLALFVTMWTDVGLCKKRPKPGGWNTGGSRYPGQGSPGGNRYPPQGGTWGQPH  60

Query  61   hgggwgqphgggwgqphgggwgqphgggwgqggg THSQWNKPSKPKTNMKHMagaaaaga  120
             GG      G       G       GG    GG TH+QWNKPSKPKTN+KH+AGAAAAGA
Sbjct  61   GGGWGQPHGGSWGQPHGGSWGQPHGGGWGQGGG-THNQWNKPSKPKTNLKHVAGAAAAGA  119

Query  121  vvgglggymlgsamsRPIIHFGSDYEDRYYRENMHRYPNQVYYRPMDEYSNQNNFVHDCV  180
            VVGGLGGYMLGSAMSRP+IHFG+D+EDRYYRENM+RYPNQVYYRP+D+YSNQNNFVHDCV
Sbjct  120  VVGGLGGYMLGSAMSRPMIHFGNDWEDRYYRENMYRYPNQVYYRPVDQYSNQNNFVHDCV  179

Query  181  NITIKQHtvttttkgenftetDVKMMERVVEQMCITQYERESQAYY--QRGSSMVLFS  236
            NITIKQHTVTTTTKGENFTETDVKMMERVVEQMC+TQY++ESQAYY  +R SS VLFS
Sbjct  180  NITIKQHTVTTTTKGENFTETDVKMMERVVEQMCVTQYQKESQAYYDGRRSSSTVLFS  237
```

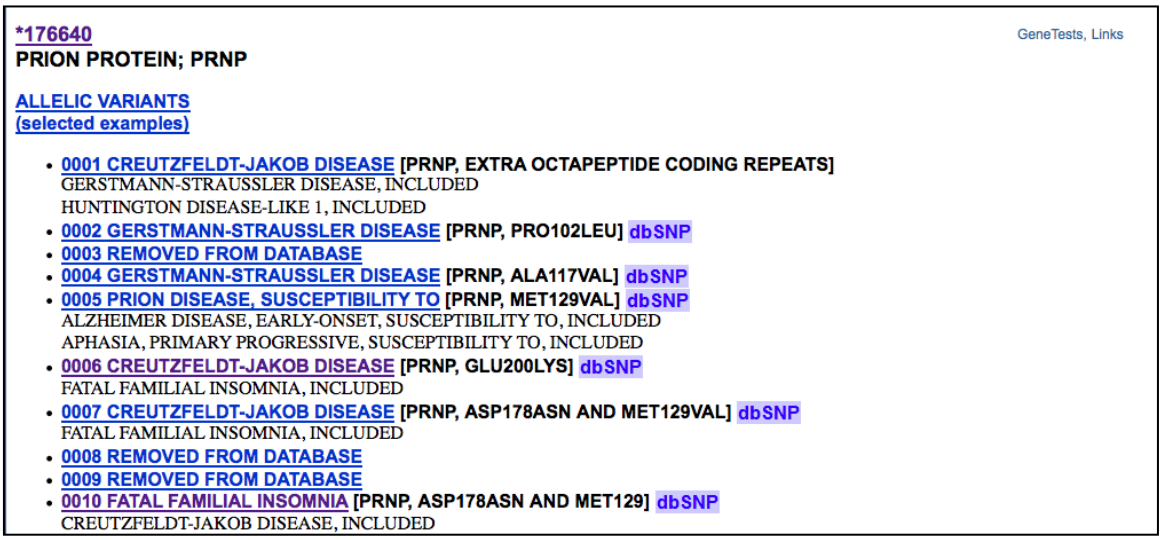

Genomic regions, transcripts, and products
(plus) Go to reference sequence details
Try our new Sequence Viewer
NC_000020.10
Genomic context
chromosome: 20; Location: 20p13
See PRNP in MapViewer
Bibliography
GEO Profiles
Genome
HomoloGene
Map Viewer
Nucleotide
OMIM
Probe
Protein
PubChem Compound
PubChem Substance
PubMed
PubMed (GeneRIF)
PubMed (OMIM)
RefSeq Proteins
RefSeq RNAs
RefSeqGene
SNP
SNP: GeneView
SNP: Genotype
SNP: VarView
Taxonomy
UniSTS
UniGene
LinkOut

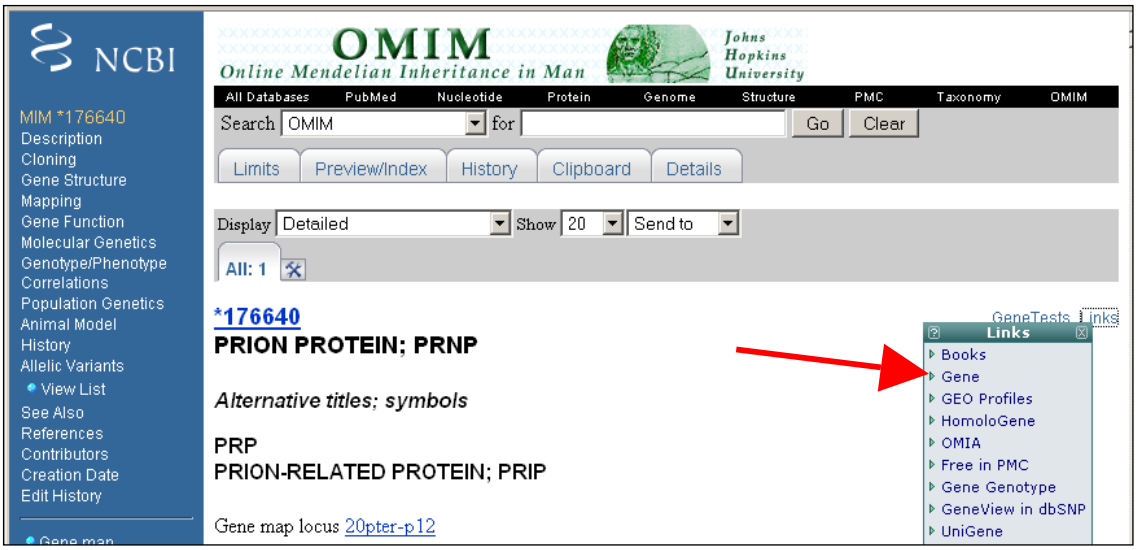

NCBI
ENTREZ SNP
Single Nucleotide Polymorphism
All Databases PubMed Nucleotide Protein Genome Structure OMIM PMC Journals Books
Search SNP for Go Clear
Limits Preview/Index History Clipboard Details
Display Graphic Summary Show 20 Sort By Send to
All: 184 1000 Genomes: 73 Cited in PubMed: 8 Clinical/LSDB Submissions: 20 Human: 184
Items 1 - 20 of 184
Page 1 of 10 Next
1: rs80309636 [Homo sapiens]
Links
AATATTAATCTCAATCCAGGTTAAGC[A/T]AAATTTTTTGCTCTCCTCTTTAGAA
MapView No VarVu No PubMed GeneView Not on mRNA No 3D No OMIM
HGVS Names: [NT_011387.8:g.4606663A>T]
2: rs80282372 [Homo sapiens]
Links
TGGGGGGCTGGGGGAGCTGGCGGAGC[A/C]CGTTAGGGAGGTCGGTGGCGCCGGG
MapView No VarVu No PubMed GeneView SeqView No 3D No OMIM
HGVS Names: [NM_000311.3:c.-11+278C>A] [NM_001080121.1:c.-6+278C>A] [NM_001080122.1:c.-6+282C>A] [NM_001080123.1:c.-11+54C>A] [NM_183079.2:c.-11+282C>A] [NT_011387.8:g.4607436C>A]
3: rs80166482 [Homo sapiens]
Links

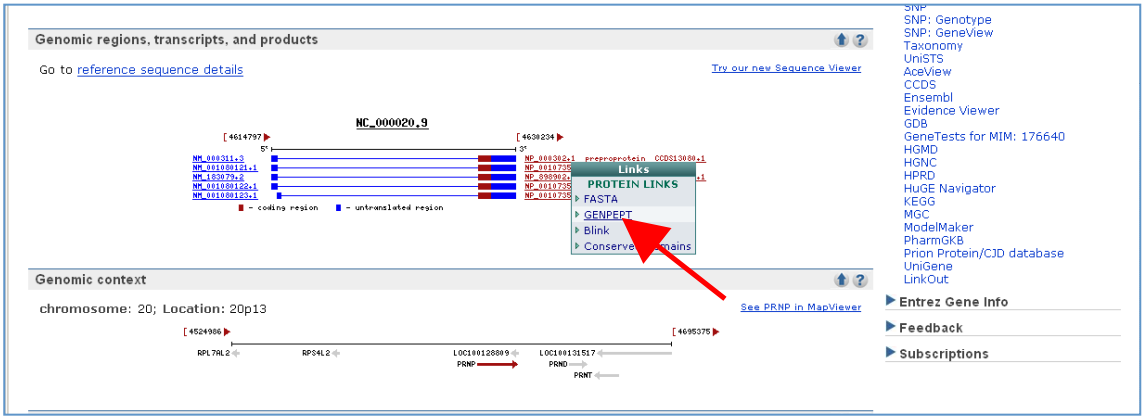

NCBI
ENTREZ SNP
Single Nucleotide Polymorphism
All Databases PubMed Nucleotide Protein Genome Structure OMIM PMC Journals Books
Search SNP for Go Clear
Limits Preview/Index History Clipboard Details
Display Graphic Summary Show 20 Sort By Send to
All: 184 1000 Genomes: 73 Cited in PubMed: 8 Clinical/LSDB Submissions: 20 Human: 184
Items 1 - 20 of 20
One page.
1: rs74315415 [Homo sapiens]
Links
CCTTGGCGGCTACATGCTGGGAAGTG[C/T]CATGAGCAGGCCCATCATACATTTC
Un no Map VarView No PubMed No Gene Not on mRNA No 3D OMIM
2: rs74315414 [Homo sapiens]
Links
ACAGTCAGTGGAACAAGCCGAGTAAG[A/C]CAAAAACCAACATGAAGCACATGGC
Un no Map VarView No PubMed No Gene Not on mRNA No 3D OMIM
3: rs74315413 [Homo sapiens]
Links
CTGCGTCAATATCACAATCAAGCAGC[A/G]CACGGTCACCACAACCACCAAGGGG
Un no Map VarView No PubMed No Gene Not on mRNA No 3D OMIM
4: rs74315412 [Homo sapiens]
Links
CGAGACCGACGTTAAGATGATGGAGC[A/G]CGTGGTTGAGCAGATGTGTATCACC
Un no Map VarView No PubMed No Gene Not on mRNA No 3D OMIM
5: rs74315411 [Homo sapiens]
Links

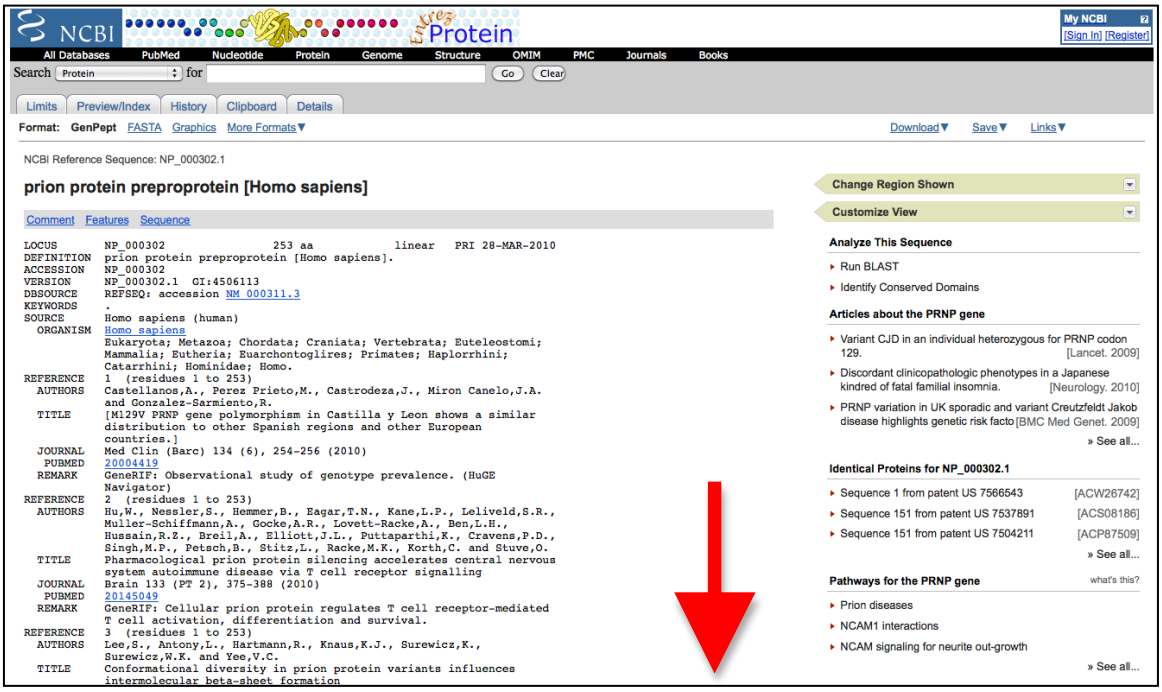

Un no Map VarView No PubMed No Gene Not on mRNA No 3D OMIM
16: rs28933385 [Homo sapiens]
Links
CAACCACCAAGGGGGAGAACTTCACC[A/G]AGACCGACGTTAAGATGATGGAGCG
20 MapView VarView No PubMed No Gene Not on mRNA No 3D OMIM
HGVS Names: [NG_009087.1:g.18668G>A] [NM_000311.3:c.598G>A] [NM_001080121.1:c.598G>A] [NM_001080122.1:c.598G>A] [NM_001080123.1:c.598G>A] [NM_183079.2:c.598G>A] [NP_000302.1:p.Glu200Lys] [NP_001073590.1:p.Glu200Lys] [NP_001073591.1:p.Glu200Lys] [NP_001073592.1:p.Glu200Lys] [NP_898902.1:p.Glu200Lys] [NT_011387.8:g.4620464G>A]
17: rs16990018 [Homo sapiens]
Links
CTACAGGCCCATGGATGAGTACAGCA[A/G]CCAGAACAACTTTGTGCACGACTGC
20 MapView VarView No PubMed No Gene Not on mRNA No 3D OMIM
HGVS Names: [NG_009087.1:g.18582A>G] [NM_000311.3:c.512A>G] [NM_001080121.1:c.512A>G] [NM_001080122.1:c.512A>G] [NM_001080123.1:c.512A>G] [NM_183079.2:c.512A>G] [NP_000302.1:p.Asn171Ser] [NP_001073590.1:p.Asn171Ser] [NP_001073591.1:p.Asn171Ser] [NP_001073592.1:p.Asn171Ser] [NP_898902.1:p.Asn171Ser] [NT_011387.8:g.4620378A>G]
18: rs11538758 [Homo sapiens]
Links
CGAP-GAI
CAGTCAGTGGAACAAGCCGAGTAAGC[A/C/T]AAAAACCAACATGAAGCACATGGCT
20 MapView VarView No PubMed No Gene Not on mRNA No 3D OMIM
HGVS Names: [NG_009087.1:g.18384C>A] [NG_009087.1:g.18384C>T] [NM_000311.3:c.314C>A] [NM_000311.3:c.314C>T] [NM_001080121.1:c.314C>A] [NM_001080121.1:c.314C>T] [NM_001080122.1:c.314C>A] [NM_001080122.1:c.314C>T] [NM_001080123.1:c.314C>A] [NM_001080123.1:c.314C>T] [NM_183079.2:c.314C>A] [NM_183079.2:c.314C>T] [NP_000302.1:p.Pro105Gln] [NP_000302.1:p.Pro105Leu] [NP_001073590.1:p.Pro105Gln] [NP_001073590.1:p.Pro105Leu] [NP_001073591.1:p.Pro105Gln] [NP_001073591.1:p.Pro105Leu] [NP_001073592.1:p.Pro105Gln] [NP_001073592.1:p.Pro105Leu] [NP_898902.1:p.Pro105Gln] [NP_898902.1:p.Pro105Leu] [NT_011387.8:g.4620180C>A] [NT_011387.8:g.4620180C>T]

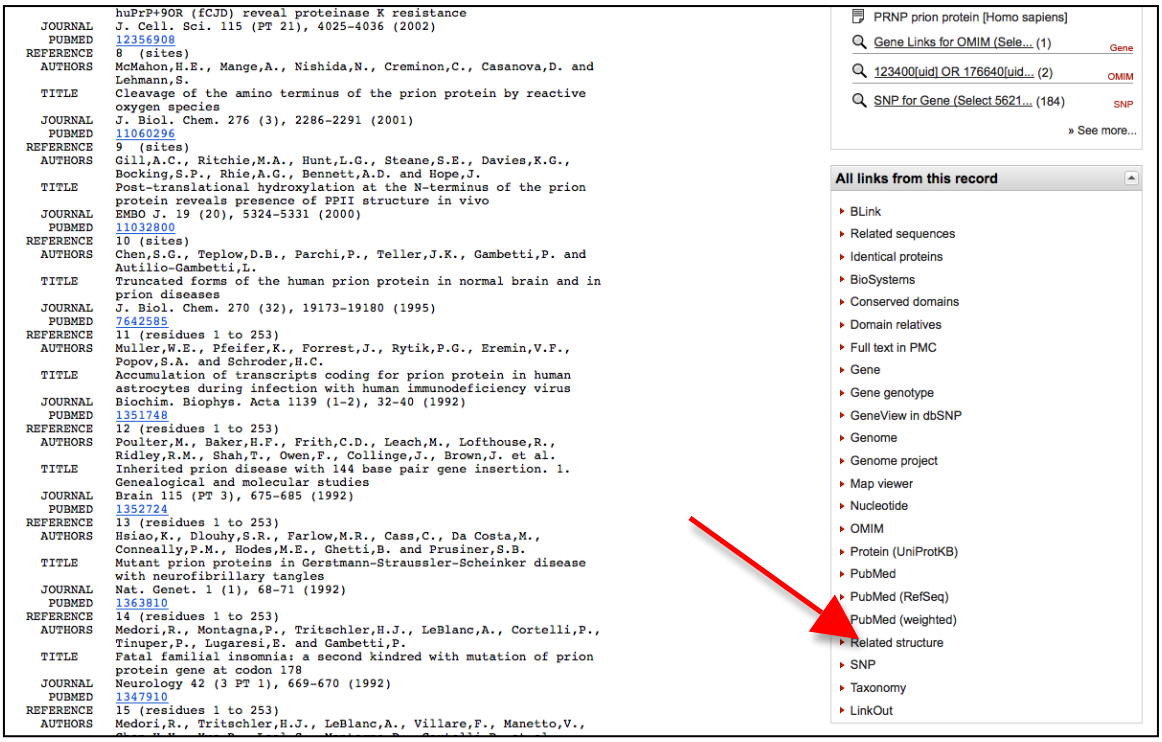

NCBI
OMIM
Online Mendelian Inheritance in Man
Johns Hopkins University
My NCBI
[Sign In] [Register]
All Databases PubMed Nucleotide Protein Genome Structure PMC Taxonomy OMIM
Search OMIM for Go Clear
Limits Preview/Index History Clipboard Details
Display Detailed Show 20 Send to
MIM *176640
Description
Cloning
Gene Structure
Mapping
Gene Function
Molecular Genetics
Genotype/Phenotype Correlations
Population Genetics
Animal Model
History
Allelic Variants
View List
See Also
References
Contributors
Creation Date
Edit History
Gene map
Entrez Gene
Nomenclature
RefSeq
GenBank
Protein
UniGene
LinkOut
HGVS
HGMD
OAD
*176640
GeneTests, Links
PRION PROTEIN; PRNP
Alternative titles; symbols
PRP
PRION-RELATED PROTEIN; PRIP
Gene map locus 20pter-p12
TEXT
DESCRIPTION
The PRNP gene encodes the prion protein, which has been implicated in various types of transmissible neurodegenerative spongiform encephalopathies. The human prion diseases occur in inherited, acquired, and sporadic forms. Approximately 15% are inherited and associated with coding mutations in the PRNP gene. Inherited prion diseases include familial Creutzfeldt-Jakob disease (CJD; 123400), Gerstmann-Straussler disease (GSD; 137440), and fatal familial insomnia (FFI; 600072). Acquired prion diseases include iatrogenic CJD, kuru (245300), variant CJD (vCJD) in humans, scrapie in sheep, and bovine spongiform encephalopathy (BSE) in cattle. Prion diseases are also referred to as transmissible spongiform encephalopathies (TSE). Variant CJD is believed to be acquired from cattle infected with BSE. However, the majority of human cases of prion disease occur as sporadic CJD (sCJD) (Collinge et al., 1996; Parchi et al., 2000; Hill et al., 2003).
CLONING
Oesch et al. (1985) isolated a cDNA clone corresponding to a pathogenic PrP fragment from a scrapie-infected hamster brain cDNA library. Southern blotting with PrP cDNA revealed a single gene with the same restriction patterns in normal and scrapie-infected brain DNA. A single PrP-related gene was also detected in murine and human DNA. Proteinase K digestion yielded PrP 27-30 in infected brain extract, but completely degraded the PrP-related protein in normal brain extract.
Kretzschmar et al. (1986) isolated a PRNP cDNA from a human retina cDNA library. The 253-amino acid protein shared 90% amino acid sequence identity with the hamster protein. Northern blot analysis detected a 2.5-kb mRNA in a variety of human neuroectodermal cell lines.
Basler et al. (1986) determined that the pathogenic PrP protein in scrapie and normal cellular PrP are encoded by the same gene. The PrP coding sequence encodes an amino-termina signal peptide. The primary structure of PrP encoded by the gene of a healthy animal did not differ from that encoded by a cDNA from a scrapie-infected animal, suggesting that the

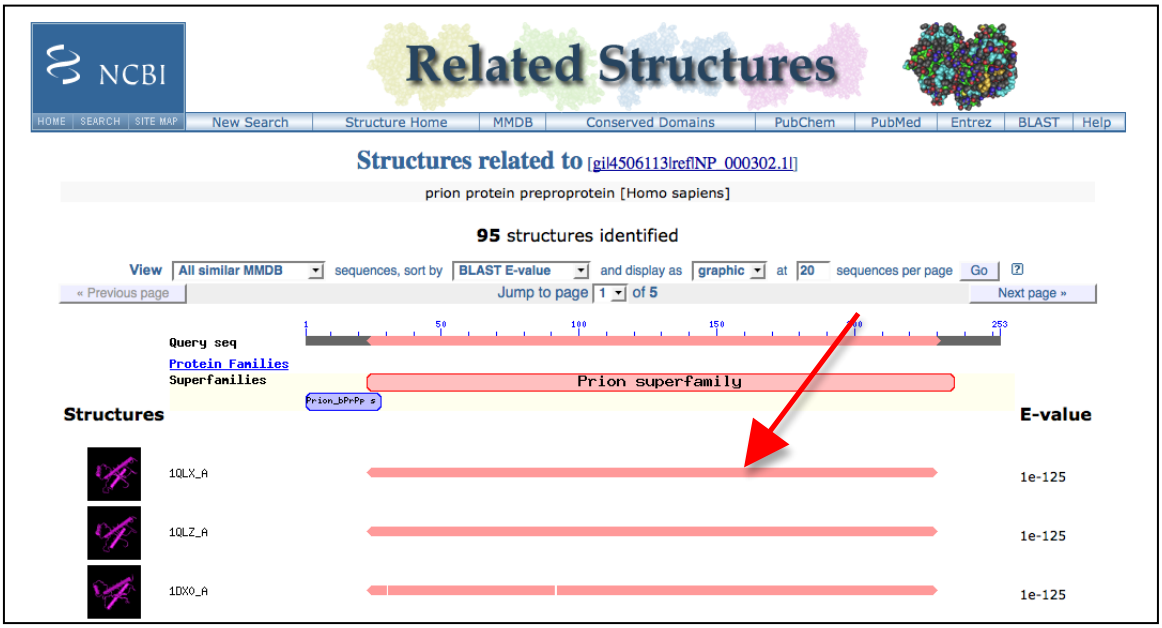

*176640
GeneTests, Links
PRION PROTEIN; PRNP
ALLELIC VARIANTS
(selected examples)
0001 CREUTZFELDT-JAKOB DISEASE [PRNP, EXTRA OCTAPEPTIDE CODING REPEATS]
GERSTMANN-STRAUSSLER DISEASE, INCLUDED
HUNTINGTON DISEASE-LIKE 1, INCLUDED
0002 GERSTMANN-STRAUSSLER DISEASE [PRNP, PRO102LEU] dbSNP
0003 REMOVED FROM DATABASE
0004 GERSTMANN-STRAUSSLER DISEASE [PRNP, ALA117VAL] dbSNP
0005 PRION DISEASE, SUSCEPTIBILITY TO [PRNP, MET129VAL] dbSNP
ALZHEIMER DISEASE, EARLY-ONSET, SUSCEPTIBILITY TO, INCLUDED
APHASIA, PRIMARY PROGRESSIVE, SUSCEPTIBILITY TO, INCLUDED
0006 CREUTZFELDT-JAKOB DISEASE [PRNP, GLU200LYS] dbSNP
FATAL FAMILIAL INSOMNIA, INCLUDED
0007 CREUTZFELDT-JAKOB DISEASE [PRNP, ASP178ASN AND MET129VAL] dbSNP
FATAL FAMILIAL INSOMNIA, INCLUDED
0008 REMOVED FROM DATABASE
0009 REMOVED FROM DATABASE
0010 FATAL FAMILIAL INSOMNIA [PRNP, ASP178ASN AND MET129] dbSNP
CREUTZFELDT-JAKOB DISEASE, INCLUDED

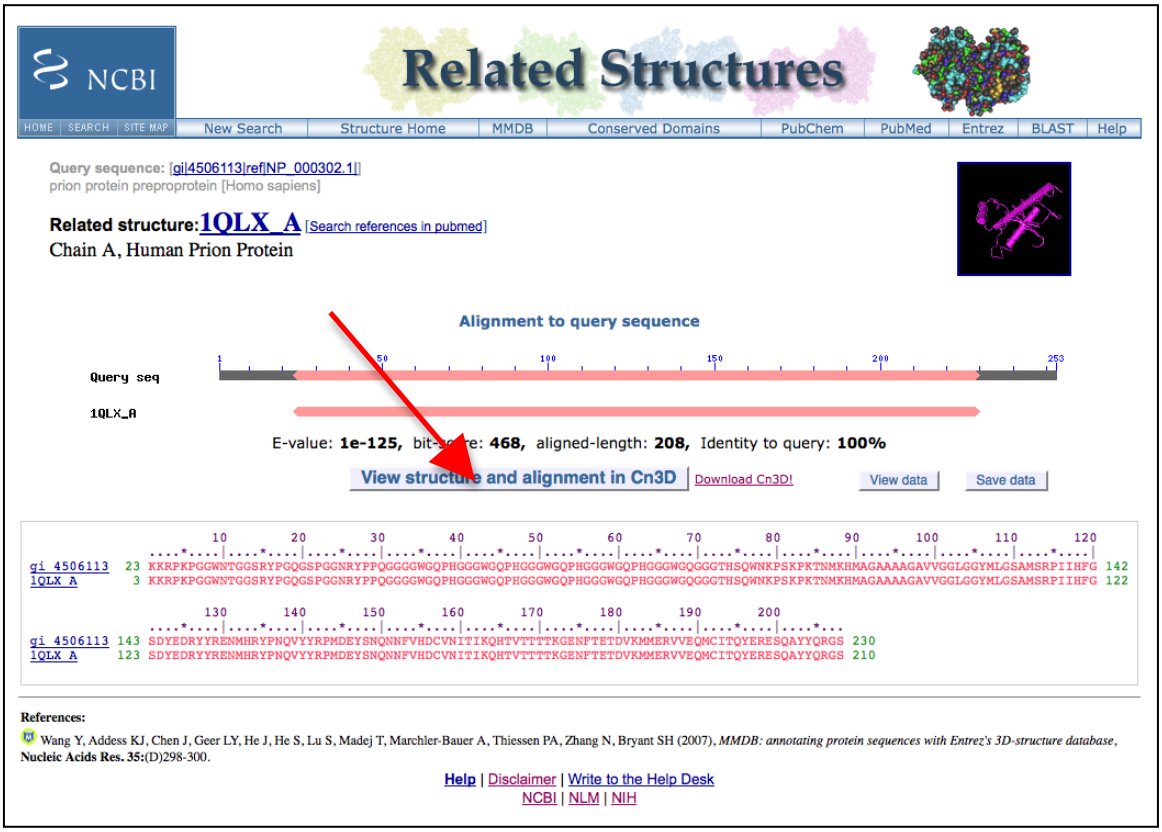

NCBI
OMIM
Online Mendelian Inheritance in Man
Johns Hopkins University
All Databases PubMed Nucleotide Protein Genome Structure PMC Taxonomy OMIM
Search OMIM for Go Clear
Limits Preview/Index History Clipboard Details
Display Detailed Show 20 Send to
All: 1
MIM *176640
Description
Cloning
Gene Structure
Mapping
Gene Function
Molecular Genetics
Genotype/Phenotype
Correlations
Population Genetics
Animal Model
History
Allelic Variants
View List
See Also
References
Contributors
Creation Date
Edit History
*176640
PRION PROTEIN; PRNP
Alternative titles; symbols
PRP
PRION-RELATED PROTEIN; PRIP
Gene map locus 20pter-p12
GeneTests Links
Links
Books
Gene
GEO Profiles
HomoloGene
OMIA
Free in PMC
Gene Genotype
GeneView in dbSNP
UniGene

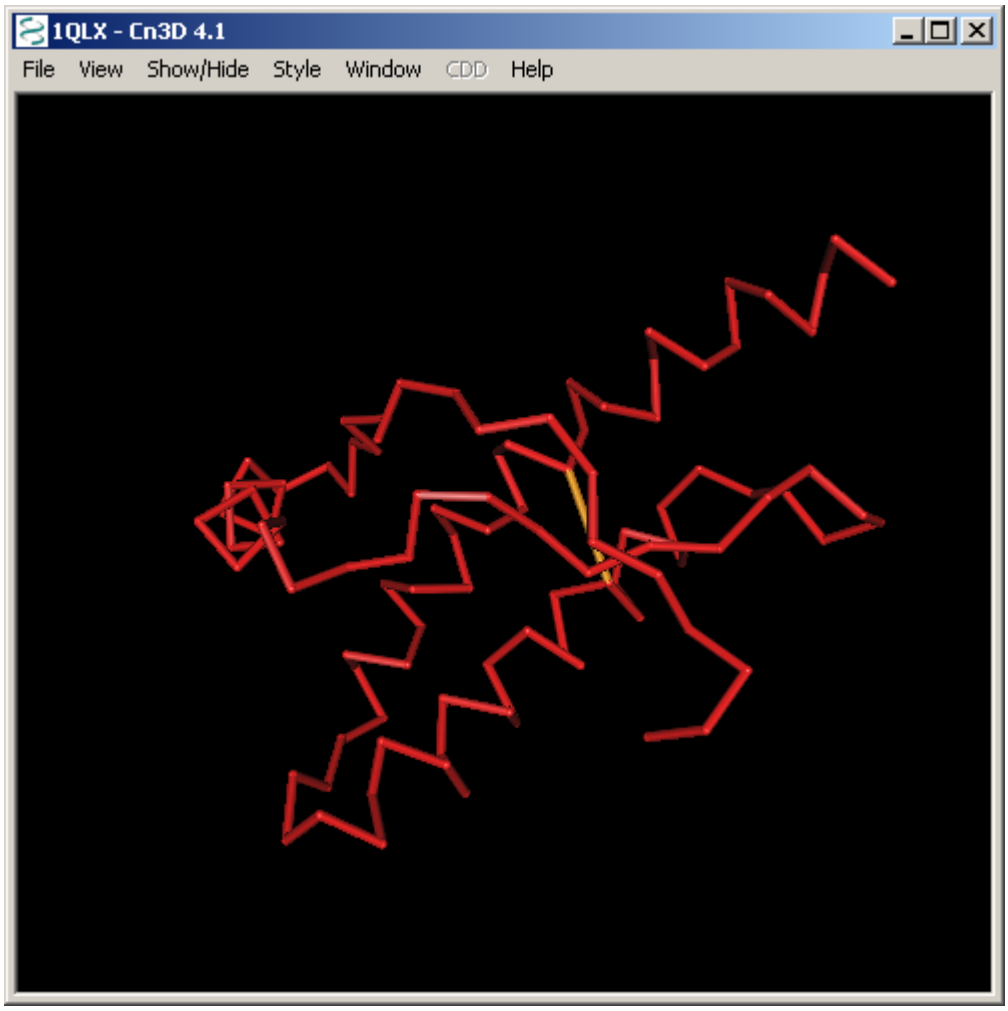

Genomic regions, transcripts, and products
Go to reference sequence details
Try our new Sequence Viewer
NC_000020.9
Links
PROTEIN LINKS
FASTA
GENPEPT
Blink
Genomic context
chromosome: 20; Location: 20p13
See PRNP in MapViewer
SNP
SNP: Genotype
SNP: GeneView
Taxonomy
UniSTS
AceView
CCDS
Ensembl
Evidence Viewer
GDB
GeneTests for MIM: 176640
HGMD
HGNC
HPRD
HuGE Navigator
KEGG
MGC
ModelMaker
PharmGKB
Prion Protein/CJD database
UniGene
LinkOut
Entrez Gene Info
Feedback
Subscriptions

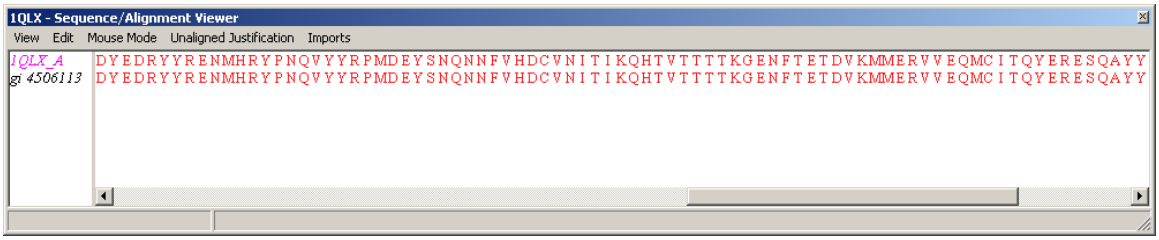

NCBI
Protein
My NCBI
[Sign In] [Register]
All Databases PubMed Nucleotide Protein Genome Structure OMIM PMC Journals Books
Search Protein for Go Clear
Limits Preview/Index History Clipboard Details
Format: GenPept FASTA Graphics More Formats▼
Download▼ Save▼ Links▼
NCBI Reference Sequence: NP_000302.1
prion protein preproprotein [Homo sapiens]
Comment Features Sequence
LOCUS NP_000302 253 aa linear PRI 28-MAR-2010
DEFINITION prion protein preproprotein [Homo sapiens].
ACCESSION NP_000302
VERSION NP_000302.1 GI:4506113
DBSOURCE REFSEQ: accession NM_000311.3
KEYWORDS .
SOURCE Homo sapiens (human)
ORGANISM Homo sapiens
Eukaryota; Metazoa; Chordata; Craniata; Vertebrata; Euteleostomi;
Mammalia; Eutheria; Euarchontoglires; Primates; Haplorrhini;
Catarrhini; Hominidae; Homo.
REFERENCE 1 (residues 1 to 253)
AUTHORS Castellanos,A., Perez Prieto,M., Castrodeza,J., Miron Canelo,J.A.
and Gonzalez-Sarmiento,R.
TITLE [M129V PRNP gene polymorphism in Castilla y Leon shows a similar
distribution to other Spanish regions and other European
countries.]
JOURNAL Med Clin (Barc) 134 (6), 254-256 (2010)
PUBMED 20004419
REMARK GeneRIF: Observational study of genotype prevalence. (HuGE
Navigator)
REFERENCE 2 (residues 1 to 253)
AUTHORS Hu,W., Nessler,S., Hemmer,B., Eagar,T.N., Kane,L.P., Leliveld,S.R.,
Muller-Schiffmann,A., Gocke,A.R., Lovett-Racke,A., Ben,L.H.,
Hussain,R.Z., Breil,A., Elliott,J.L., Puttaparthi,K., Cravens,P.D.,
Singh,M.P., Petzsch,B., Stitz,L., Racke,M.K., Korth,C. and Stuve,O.
TITLE Pharmacological prion protein silencing accelerates central nervous
system autoimmune disease via T cell receptor signalling
JOURNAL Brain 133 (PT 2), 375-388 (2010)
PUBMED 20145049
REMARK GeneRIF: Cellular prion protein regulates T cell receptor-mediated
T cell activation, differentiation and survival.
REFERENCE 3 (residues 1 to 253)
AUTHORS Lee,S., Antony,L., Hartmann,R., Knaus,K.J., Surewicz,K.,
Surewicz,W.K. and Yee,V.C.
TITLE Conformational diversity in prion protein variants influences
intermolecular beta-sheet formation
Change Region Shown
Customize View
Analyze This Sequence
Run BLAST
Identify Conserved Domains
Articles about the PRNP gene
Variant CJD in an individual heterozygous for PRNP codon 129. [Lancet. 2009]
Discordant clinicopathologic phenotypes in a Japanese kindred of fatal familial insomnia. [Neurology. 2010]
PRNP variation in UK sporadic and variant Creutzfeldt Jakob disease highlights genetic risk facto [BMC Med Genet. 2009]
» See all...
Identical Proteins for NP_000302.1
Sequence 1 from patent US 7566543 [ACW26742]
Sequence 151 from patent US 7537891 [ACS08186]
Sequence 151 from patent US 7504211 [ACP87509]
» See all...
Pathways for the PRNP gene what's this?
Prion diseases
NCAM1 interactions
NCAM signaling for neurite out-growth
» See all...


huPrP+90R (fCJD) reveal proteinase K resistance
JOURNAL J. Cell. Sci. 115 (PT 21), 4025-4036 (2002)
PUBMED 12356908
REFERENCE 8 (sites)
AUTHORS McMahon,H.E., Mange,A., Nishida,N., Creminon,C., Casanova,D. and
Lehmann,S.
TITLE Cleavage of the amino terminus of the prion protein by reactive
oxygen species
JOURNAL J. Biol. Chem. 276 (3), 2286-2291 (2001)
PUBMED 11060296
REFERENCE 9 (sites)
AUTHORS Gill,A.C., Ritchie,M.A., Hunt,L.G., Steane,S.E., Davies,K.G.,
Bocking,S.P., Rhie,A.G., Bennett,A.D. and Hope,J.
TITLE Post-translational hydroxylation at the N-terminus of the prion
protein reveals presence of PPII1 structure in vivo
JOURNAL EMBO J. 19 (20), 5324-5331 (2000)
PUBMED 11032800
REFERENCE 10 (sites)
AUTHORS Chen,S.G., Teplow,D.B., Parchi,P., Teller,J.K., Gambetti,P. and
Autilio-Gambetti,L.
TITLE Truncated forms of the human prion protein in normal brain and in
prion diseases
JOURNAL J. Biol. Chem. 270 (32), 19173-19180 (1995)
PUBMED 7642585
REFERENCE 11 (residues 1 to 253)
AUTHORS Muller,W.E., Pfeifer,K., Forrest,J., Rytik,P.G., Eremin,V.F.,
Popov,S.A. and Schroder,H.C.
TITLE Accumulation of transcripts coding for prion protein in human
astrocytes during infection with human immunodeficiency virus
JOURNAL Biochim. Biophys. Acta 1139 (1-2), 32-40 (1992)
PUBMED 1351748
REFERENCE 12 (residues 1 to 253)
AUTHORS Poulter,M., Baker,H.F., Frith,C.D., Leach,M., Lofthouse,R.,
Ridley,R.M., Shah,T., Owen,F., Collinge,J., Brown,J. et al.
TITLE Inherited prion disease with 144 base pair gene insertion. 1.
Genealogical and molecular studies
JOURNAL Brain 115 (PT 3), 675-685 (1992)
PUBMED 1352724
REFERENCE 13 (residues 1 to 253)
AUTHORS Hsiao,K., Dlouhy,S.R., Farlow,M.R., Cass,C., Da Costa,M.,
Conneally,P.M., Hodes,M.E., Ghetti,B. and Prusiner,S.B.
TITLE Mutant prion proteins in Gerstmann-Straussler-Scheinker disease
with neurofibrillary tangles
JOURNAL Nat. Genet. 1 (1), 68-71 (1992)
PUBMED 1363810
REFERENCE 14 (residues 1 to 253)
AUTHORS Medori,R., Montagna,P., Tritschler,H.J., LeBlanc,A., Cortelli,P.,
Tinuper,P., Lugaresi,E. and Gambetti,P.
TITLE Fatal familial insomnia: a second kindred with mutation of prion
protein gene at codon 178
JOURNAL Neurology 42 (3 PT 1), 669-670 (1992)
PUBMED 1347910
REFERENCE 15 (residues 1 to 253)
AUTHORS Medori,R., Tritschler,H.J., LeBlanc,A., Villare,F., Manetto,V.,
PRNP prion protein [Homo sapiens]
Gene Links for OMIM (Sele... (1) Gene
123400[uid] OR 176640[uid... (2) OMIM
SNP for Gene (Select 5621... (184) SNP
» See more...
All links from this record
BLink
Related sequences
Identical proteins
BioSystems
Conserved domains
Domain relatives
Full text in PMC
Gene
Gene genotype
GeneView in dbSNP
Genome
Genome project
Map viewer
Nucleotide
OMIM
Protein (UniProtKB)
PubMed
PubMed (RefSeq)
PubMed (weighted)
Related structure
SNP
Taxonomy
LinkOut


NCBI
Related Structures
HOME SEARCH SITE MAP
New Search
Structure Home
MMDB
Conserved Domains
PubChem
PubMed
Entrez
BLAST
Help
Structures related to [gi|4506113|ref|NP_000302.1|]
prion protein preproprotein [Homo sapiens]
95 structures identified
View All similar MMDB sequences, sort by BLAST E-value and display as graphic at 20 sequences per page Go
« Previous page
Jump to page 1 of 5
Next page »
Query seq
Protein Families
Superfamilies
Prion superfamily
Prion_bPrPp_s
Structures
E-value
1QLX_A
1e-125
1QLZ_A
1e-125
1DX0_A
1e-125


NCBI
Related Structures
HOME SEARCH SITE MAP
New Search
Structure Home
MMDB
Conserved Domains
PubChem
PubMed
Entrez
BLAST
Help
Query sequence: [gi|4506113|ref|NP_000302.1|]
prion protein preproprotein [Homo sapiens]
Related structure: 1QLX_A [Search references in pubmed]
Chain A, Human Prion Protein
Alignment to query sequence
Query seq
1QLX_A
E-value: 1e-125, bit-score: 468, aligned-length: 208, Identity to query: 100%
View structure and alignment in Cn3D
Download Cn3D!
View data
Save data
gi 4506113 23 KKRPKPGGWNTGGSRYPGQGSPGGNRYPPQGGGGWGQPHGGGWGQPHGGGWGQPHGGGWGQPHGGGWGQGGGTHSQWNKPSKPKTNMKHMAGAAAAGAVVGGLGGYMLGSAMSRPIIHFG 142
1QLX_A 3 KKRPKPGGWNTGGSRYPGQGSPGGNRYPPQGGGGWGQPHGGGWGQPHGGGWGQPHGGGWGQPHGGGWGQGGGTHSQWNKPSKPKTNMKHMAGAAAAGAVVGGLGGYMLGSAMSRPIIHFG 122
gi 4506113 143 SDYEDRYYRENMHRYPNQVYYRPMDEYSNQNNFVHDCVNITIKQHTVTTTTKGENFTETDVKMMERVVEQMCITQYERESQAYYQRGS 230
1QLX_A 123 SDYEDRYYRENMHRYPNQVYYRPMDEYSNQNNFVHDCVNITIKQHTVTTTTKGENFTETDVKMMERVVEQMCITQYERESQAYYQRGS 210
References:
Wang Y, Addess KJ, Chen J, Geer LY, He J, He S, Lu S, Madej T, Marchler-Bauer A, Thiessen PA, Zhang N, Bryant SH (2007), MMDB: annotating protein sequences with Entrez's 3D-structure database, Nucleic Acids Res. 35:(D)298-300.
Help | Disclaimer | Write to the Help Desk
NCBI | NLM | NIH





Exercise 2

Retrieve human entries related to "colon cancer" in Entrez Gene. Identify the gene MLH1. Name the map location of this gene on the human genome. What is the function of this protein? What are the alternate gene symbols? Name the phenotypes associated with the mutations in this gene.

Is the RefSeq mRNA record reviewed? How many alternatively spliced products have been annotated for the gene?

To obtain information about the homologs from other eukaryotes, click on the Homologene link. Change the Display option to "Alignment Scores". How great is the percent identity between the human and mouse proteins? View the alignment by clicking on the "Blast" link.

Go back to the Entrez Gene report. Identify the variations annotated on this gene by clicking on the geneView in dbSNP link. How many of them are missense (non-synonymous) changes? To determine whether known SNPs in the coding region of a gene are associated with any phenotype, access the OMIM record by clicking on the "Yes" link under the OMIM column in the SNP report. Compare the non-synonymous changes from the SNP report with the "ALLELIC VARIANTS" in the OMIM record. Are there any SNPs known to cause a change in the function of the MLH1 protein?

Go back to the Entrez gene report. To view the site of mutation in the 3D structure, superimpose the protein sequence on the 3D-structure of E.coli multL protein. Select the GENPEPT link for NP_000240 under the section “Genomic Region, Transcripts and products”. Then select “Related Structure” from the Links menu, click on the first arrow representing the related structure and then on the “Get 3D-structure data” button. Identify and highlight the amino acid corresponding to the human MLH1 isoleucine 32 on the 3D structure. What is the amino acid at this position in the E.coli protein? Based on this information, do you think the I32V mutation in the human protein will alter its function?